LIVING WITH DIFFERENCES IN SEX DEVELOPMENT/INTERSEX

Disclosure, sexual health, perspectives on surgery and stigma

Line Merete Mediå

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This thesis is dedicated to my mother

Line Merete Mediå

Oslo, February, 2023

Abbreviations and Clarification of Concepts

AIS - Androgen insensitivity syndrome

AYAs - Adolescents and young adults

CAH – Congenital adrenal hypoplasia

CAIS - Complete androgen insensitivity syndrome

DSD – Differences of sex development

DSD - related surgery: Encompasses surgical interventions directed at the gonads to reduce risk of malignancy, internal reproductive structures, or external genitalia (Gardner & Sandberg, 2018)

FGI – Focus group interview

HCP – Health care professionals

HRT - Hormone replacement therapy

HUS – Haukeland University Hospital

LGBTI - Organizations for lesbian, gay, bisexual, transgender, and intersex

MRKH - Mayer-Rokitansky-Küster-Hauser syndrome

OUS – Oslo University Hospital

PAIS - Partial androgen insensitivity syndrome

Phenotype – refers to an individual's visible, or observable traits such as eye color or height. Phenotypic sex can be less visible as it "refers to an individual's sex as determined by their internal and external genitalia, expression of secondary sex characteristics, and behavior" (Purves et al., 2001).

QoL - Quality of life

RTA - Reflexive thematic analysis

SDM - Shared decision making

TS - Turner syndrome

Summary

Background

Differences in sex development (DSD)/intersex are rare conditions that affect individuals' sex development in different ways, causing their genitals, hormones, and/or chromosomes to differ from traditional conceptions of male and female bodies. Moreover, DSD/intersex comprise a group of conditions that are diverse with regard to the associated diagnoses, severity of medical complications, psychological impacts, treatments, and follow-up.

Aim

This thesis explores the experiences of individuals with DSD/intersex conditions as well as the dilemmas faced by health-care professionals (HCPs) who work with individuals born with DSD/intersex. Issues such as everyday challenges, disclosure, information sharing, and stigma are explored, as are the dilemmas related to decisions concerning DSD/intersex-related surgery.

Materials and methods

The empirical material in this thesis is derived from individual in-depth qualitative interviews conducted with 26 Norwegian participants who were all born with a DSD/intersex condition and from three focus group interviews held with 14 HCPs who work with DSD/intersex patients. Among the participants born with a DSD/intersex condition, 18 identify as female, 8 as male, and none as non-binary. As a group, the participants have seven different DSD/intersex conditions and a wide range of different backgrounds and experiences. A total of 11 participants are adolescents and young adults (AYAs) aged 16–26 years, whereas 15 participants are adults aged 30–70+ years. The participating HCPs have all worked within or in collaboration with multidisciplinary DSD/intersex teams for 1–30+ years. Eight HCPs identify as female and six as male. This thesis has a qualitative design that entails an interpretive phenomenological perspective and a reflexive thematic analysis.

Results

The findings detailed in this thesis are presented in three papers, all of which address how stigma influences the lives of individuals with DSD/intersex conditions as well as how stigma results in dilemmas when it comes to decision making concerning DSD/intersex-related surgery. Among the adults aged 30–70 years who were born with a DSD/intersex condition, the expectation of facing stigma and the feeling of not having mastered discussion

of their condition resulted in the need to strike a balance between hiding and/or exposing their condition in an effort to pass as "normal" or to achieve understanding. These issues are explored in the first paper. To achieve the necessary balance, several participants reported performing invisible work that is not necessarily perceivable to others but still affects their everyday lives. Interestingly, the fear and expectation of stigma changed over time, as the participants who were diagnosed in childhood noted that disclosure had become easier with advancing age.

Stigma and a need to be "normal" were related to sexual health in AYA aged 16-26 years. These issues are explored in the second paper. The participants revealed that the experienced differences in terms of both the function and appearance of their genitalia affect their sexual activities and intimate relations. As in the first paper, having limited knowledge of their condition and lacking everyday language with which to describe that condition complicate their relations with others. Additionally, the AYAs talked about how this affects their feeling of differentness and sense of being stigmatized. The female participants expressed ambivalent feelings regarding infertility, which influence the decision to discuss this issue with their partners. The participants wished for acceptance of their perceived differentness and effort to understand their sexual health needs.

The impact of stigma is also evident when the HCPs talk about how the parents of young children experience expectations of having a "normal" child and how this influences the decision-making process. This results in dilemmas regarding DSD/intersex-related surgeries, as the HCPs recognize decisions regarding such surgeries to be influenced by the fear of stigma and the lack of evidence-based practice. Further dilemmas are evident in terms of how best to support and communicate with caregivers, in addition to how best to address uncertainties regarding surgical results. These findings elucidate the complex considerations and challenges that HCPs face when guiding patients and caregivers through surgical decision-making processes in the context of DSD/intersex.

Despite the negative consequences of living with a DSD/intersex condition, several participants expressed satisfaction with their lives, with the information conveyed to them, and with their bodies after having learned to accept and appreciate their differentness. However, some participants expressed a degree of ambivalence, for example, regarding their acceptance of infertility, of the impacts their conditions have on their lives, and of the desire for normality.

Conclusion

The findings of this study show that several aspects of DSD/intersex are still influenced by stigma and, therefore, that there remains a need for more focus on comprehension and de-stigmatization of DSD/intersex with regard to parents, affected individuals, HCPs, and society in general. A lack of knowledge and understanding among individuals with DSD/intersex conditions can, in the worst-case scenario, lead to them avoiding contact with HCPs. To help address this issue, the present thesis contributes to raising awareness of DSD/intersex conditions.

Adults with DSD/intersex conditions currently receive little or no multidisciplinary follow up in Norway. Both the participants and HCPs acknowledge that adults with DSD/intersex are in need of continued medical care for their DSD/intersex, including mental health specialists. Given the findings of this thesis, a transition program should be introduced to ensure that dedicated HCPs are in charge of the multidisciplinary follow up of those who want or need such support as adults. Finally, more research needs to be conducted on the consequences of both performing DSD/intersex-related surgery and withholding such surgery.

Sammendrag

Bakgrunn

Variasjon i kroppslig kjønnsutvikling (DSD)/intersex er sjeldne tilstander som påvirker enkeltpersoners kjønnsutvikling på ulike måter, noe som resulterer i at deres kjønnsorganer, hormoner og/eller kromosomer avviker fra tradisjonelle oppfatninger av mannlige og kvinnelige kropper. DSD/intersex er en gruppe tilstander som representerer et mangfold av diagnoser, alvorlighetsgrad av medisinske komplikasjoner, psykologiske påvirkninger, behandlinger og oppfølging.

Mål

Målet med denne studien er å utforske opplevelsen til personer med DSD/intersex, sammen med dilemmaene som helsepersonell (HP) som jobber med DSD/intersex, står overfor. Tema som hverdagsutfordringer, åpenhet, informasjonsdeling og stigma ble utforsket, sammen med dilemmaer knyttet til beslutninger om DSD/intersex-relatert kirurgi.

Materialer og metoder

Det empiriske materialet i denne avhandlingen er basert på individuelle inngående kvalitative intervjuer med 26 norske deltakere som alle ble født med en DSD/intersex-tilstand, samt tre fokusgruppeintervjuer med HP (n = 14) som arbeider med DSD/intersex. Atten av deltakerne født med DSD/intersex identifiserte seg som kvinner, og åtte som menn. Ingen identifiserte seg som ikke-binære. Deltagerne representerte syv ulike DSD/intersex-tilstander og et mangfold av bakgrunn og erfaringer. Elleve av deltakerne var ungdom og unge voksne (AYA), i alderen 16-26 år. Fjorten deltakere var voksne, i alderen 30-70 år. Helsepersonell arbeidet alle innenfor, eller i samarbeid med tverrfaglige DSD-team og hadde ulike erfaringer med DSD fra 1 til over 30 år. Åtte HP identifiserte seg som kvinner, og seks som menn.

Studien har en kvalitativ design med en fortolkende fenomenologisk tilnærming. Det ble gjort en refleksiv tematisk analyse av datamaterialet.

Resultater

Funnene i denne studien er representert i tre artikler, som alle tar for seg hvordan stigma påvirker livene til individer med variasjon i kroppslig kjønnsutvikling/intersex, og også hvordan stigma griper inn i dilemmaer knyttet til beslutningsprosessen om kirurgi relatert til DSD/intersex. Vi lærte at voksne i alderen 30-70 år født med DSD/intersex, har en forventning om å bli stigmatisert, og de har en følelse av å ikke mestre å snakke om egen tilstand. Dette førte til at de voksne balanserte mellom å skjule eller og avsløre tilstanden sin for å kunne bli oppfattet som "normal", eller for å oppnå forståelse. Disse temaene ble

utforsket i den første artikkelen. For å oppnå denne balansegangen gjorde flere et usynlig arbeid som ikke nødvendigvis var merkbar for andre, men som likevel påvirket deltakernes hverdag. Vi lærte også at frykten for stigma endret seg over tid, da de som ble diagnostisert i barndommen uttrykte at det ble lettere å være åpen med økende alder. Imidlertid virket det som om å bli diagnostisert som voksen økte følelsen av å være annerledes og kompliserte åpenheten. For ungdom og unge voksne i alderen 16-26 år født med DSD/intersex, var stigma og behovet for å være "normal" knyttet til seksuell helse, og dette er utforsket i den andre artikkelen. Funn i denne studien avslørte at opplevd forskjell både i funksjon og utseende av genitalia påvirket seksuell aktivitet og intime relasjoner. Som i den første artikkelen, gjorde begrenset kunnskap om tilstanden og mangel på hverdags språk at relasjonene til andre var vanskelig. I tillegg snakket ungdom og unge voksne om hvordan dette påvirket deres følelse av å være annerledes og følelsen av å være stigmatisert. Kvinnelige deltakere uttrykte ambivalente følelser når det gjaldt infertilitet, noe som påvirket beslutningen om å diskutere dette emnet med partneren. Deltakerne ønsket aksept for sin opplevde annerledeshet og forsøkte å forstå sine behov knyttet til seksuell helse. Påvirkningen av stigma ble også tydelig når helsepersonell snakket om hvordan foreldre til små barn opplevde forventninger om å få et "normalt" barn, og hvordan dette påvirket beslutningsprosessen. Dette resulterte i dilemmaer knyttet til DSD-relaterte kirurgi, ettersom helsepersonell snakket om at beslutninger knyttet til DSD-relaterte kirurgi ble påvirket av frykten for stigma og mangelen på kunnskapsbasert praksis. Dilemmaer knyttet til hvordan man best kunne støtte og kommunisere med omsorgspersoner, og hvordan man skulle håndtere usikkerhet knyttet til kirurgiske resultater ble også diskutert blant helsepersonell. Dette kastet lys over de intrikate overveielsene og utfordringene som helsepersonell møter når de veileder pasienter og omsorgspersoner gjennom beslutningsprosesser knyttet til kirurgi i DSD-sammenheng. Til tross for de negative konsekvensene av å leve med DSD/intersex, uttrykte flere deltakere tilfredshet med livene sine, med informasjonen som ble formidlet, og med kroppene sine når de hadde lært å akseptere og verdsette sin annerledeshet. Imidlertid merket noen deltakere en grad av ambivalens, for eksempel når det gjaldt aksept av infertilitet, hvor stor innvirkning tilstanden hadde på livet deres, og streben etter normalitet.

Konklusjon

Studien viser generelt at flere aspekter ved DSD/intersex fortsatt er preget av stigma, og at det må være mer fokus på forståelse og avstigmatisering av DSD/intersex, både overfor foreldre, berørte individer, HP og samfunnet generelt. Manglende kunnskap og forståelse

blant berørte individer kan i verste fall føre til at de unngår kontakt med H~~C~~P. Følgelig kan denne avhandlingen bidra til å øke kunnskapen om DSD/intersex. Voksne med DSD/intersex mottar lite eller ingen tverrfaglig oppfølging. Basert på resultatene i denne avhandlingen konkluderer vi med at det bør være et overgangsprogram for å sikre at dedikerte HP har ansvaret for tverrfaglig oppfølging for de som er blitt voksne. Denne oppfølgingen kan skje kanskje fem eller ti år etter utskrivelse fra barneavdelingen, men at individuelle vurderinger blir gjort i henhold til dette. Til slutt konkluderer vi med at det må utføres mer forskning om konsekvensene av å gjennomføre DSD-relatert kirurgi og av å avstå fra denne kirurgien.

List of Papers

Paper 1:

Mediå, L. M., Fauske, L., Sigurdardottir, S., Feragen, K. J. B., Heggeli, C., & Wæhre, A. (2022). 'It was Supposed to be a Secret': a study of disclosure and stigma as experienced by adults with differences of sex development. *Health Psychology and Behavioral Medicine, 10*(1), 579-595. https://doi.org/10.1080/21642850.2022.2102018

Paper 2

Mediå, L. M., Sigurdardottir, S., Fauske, L., & Waehre, A. (2023). Understanding sexual health concerns among adolescents and young adults with differences of sex development: a qualitative study. *International Journal of Qualitative Studies on Health and Well-being, 18*(1), 2204635. https://doi.org/10.1080/17482631.2023.2204635

Paper3

Mediå, L. M., Fauske, L., Sigurdardottir, S., Feragen, K. J. B., & Wæhre, A. (2023). Dilemmas Faced by Health-Care Professionals Regarding Treatment and Differences of Sex Development: A Qualitative Study.

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Preface

When working in the Oslo University hospital as a newly graduated nurse, I became aware of the different approaches people had at the different hospital wards. “My” ward, a surgical department located on the fourth floor of the hospital for children with rare, congenital malformations in their genitals, urinary tract, and/or gastro-intestinal tract, did not receive much attention from the public. To me, children with severe illnesses in other wards seemed to receive more attention. For example, when the patients had to spend Christmas in the hospital. The ward on the floor below received donations of Christmas presents from different organizations to give to children who were admitted during the Christmas weekend. “My” children did not appear to receive much donations. Similarly, a child with an intravenous-infusion stand might have decorated it with colorful Christmas lights; however, when it came to a child who had undergone surgery for hypospadias, a congenital difference in the urethra opening in the penis, the urinary catheter is often hidden under a blanket. Moreover, I saw brave families with children suffering from e.g. cancer who told their stories in magazines or newspapers to enhance focus on research, whereas magazines or newspapers rarely or never publish stories about children born with malformations in the genitals or bowel. These observation sparked my curiosity. How does this difference in how rare conditions are met and understood by others affect the children, their parents, siblings, and extended families? Why is it that these rare conditions, which affect parts of the body we seldom talk about in public, seem to be treated differently from, for instance, cancer? The desire to answer these questions influenced my decision to join the University of Oslo and study for a multidisciplinary masters in the Faculty of Medicine within the Institute of Health and Society.

The intricate perspectives of lived experiences, together with medical perspectives and the cultural/social constructivist approach to health, illness, and disease, taught me that these questions could be approached in different ways, depending on the perspective of the questioner. I also learned how different yet equally important and complementary these different perspectives could be. For most of the 20th century, individuals living with differences in sex development (DSD)/intersex were seen and understood through medical lenses. Now, as there is still a long way to go regarding the development of practical clinical guidelines that are graded as strong (Nordenstrom et al., 2022), the medical community needs to acknowledge that prior research and understanding of DSD/intersex has mostly focused on the medical and clinical aspects, neglecting other approaches until quite recently. Therefore, the voices of individuals with DSD and their families still need to be brought forth. In

addition, the Norwegian Action Plan Against Discrimination on the Basis of Sexual Orientation, Gender Identity and Gender Expressions 2017–2020 has resulted in two reports: "Law and intersex in Norway: Challenges and opportunities" (Garland et al., 2018) and "Livssituasjonen for personer med variasjon i kroppslig kjønnsutvikling i Norge" ("Life situation of people with differences in sex development in Norway") (Feragen et al., 2019). These reports demonstrate the need to focus and develop further knowledge on DSD/intersex. Such thoughts led to the present research project titled "Living with a difference in sex development/intersex: Perspectives on vulnerable periods of life and dilemmas regarding surgery."

Introduction

Rationale and Background

The present thesis grew from a project initiated by the Norwegian Directorate for Children, Youth and Family Affairs (*Barne-, ungdoms- og familiedirektoratet* or *Bufdir* in Norwegian, which is referred to as "the Bufdir project" hereafter) in 2017. The Bufdir project, which was conducted by the Centre for Rare Disorders, Oslo University Hospital, investigated the life situations of individuals with differences in sex development (DSD)/intersex conditions as well as their need for health- and care services and supportive interventions. Kristin Feragen and Anne Wæhre, who were part of the research team behind the study that informed the present thesis, were involved in the Bufdir project. The author of this thesis (Line Mediå) also contributed to the project, including conducting interviews with parents and recruiting participants. The Bufdir project was reported on in "Livssituasjonen til personer med variasjon i kroppslig kjønnsutvikling i Norge" (in English "Life situations of individuals with differences of sex development in Norway") (Feragen et al., 2019). The findings of which revealed the need for a deeper understanding of the life situations of individuals with DSD/intersex and an exploration of the themes identified during the project. Thus, the Centre for Rare Disorders initiated the present Ph.D. research project.

This chapter provides an overview of the literature and topics relevant to the Ph.D. research. First, I elaborate on the nomenclature used in this thesis, which aids in understanding the psychosocial aspects of living with DSD/intersex conditions. Next, I situate DSD/intersex in a wider historical context, introduce the medical field of DSD/intersex, and describe the relevant conditions. Subsequently, I present the theoretical perspectives of stigma, epistemic injustice, and healing. Finally, I explain why research into the lived experiences of individuals with DSD/intersex conditions and the dilemmas associated with DSD/intersex-related surgeries is necessary.

A Brief Note on Nomenclature

Describing or naming diversity in terms of human sexual development is complex, and I acknowledge that sensitivity is required in this regard. There is ongoing debate concerning the nomenclature appropriate for DSD/intersex. In 2005, human right activists, parents, those with DSD/intersex conditions, and clinicians agreed on a new consensus statement regarding DSD/intersex (Lee et al., 2006). This consensus statement was intended to improve the management of DSD/intersex disorders and to ensure good clinical practice involving a new treatment model/protocol and new nomenclature that shifted from terms perceived as stressful

and stigmatizing, such as intersex and hermaphrodite, to the more medicalized "disorder of sex development" (Lee et al., 2006). Many regarded the new term "disorder of sex development" as an important step toward a more international, united approach to the treatment and follow up of children, adolescents, and adults with DSD/intersex. Another argument for using the term "disorder" was the associated shift in focus from identity, gender, sex, and genitals to issues that could be treated medically, for example, hormonal disturbances (Feder & Karkazis, 2008). However, criticism of the term has also been offered. Some consider it to be pathologizing, thereby posing the risk of medicalization and stigmatization due to the focus on the sex development being "disordered." Researchers such as Liao and Roen (2014) have advocated for the use of more inclusive and depathologizing terms, for example, "diverse sex development," "diversity in sex development," or "difference in sex development," that respect the need for health services but seek to maintain a critical distance from the medical terminology of disorders of sex development. Thus, various terms have been used to describe DSD/intersex. Here, the medical field's need for functional descriptions and classifications, on the one hand, and affected persons' need for descriptions that are not pejorative or for precise diagnostic names, on the other hand, must be taken into account (Kim & Kim, 2012).

In the Bufdir project, most participants considered diversity of sex development to be the most descriptive nomenclature for their condition (Feragen et al., 2019). However, a few referred to disorder of sex development, while most just used the name of the relevant condition (e.g., Klinefelter syndrome or congenital adrenal hyperplasia). Some individuals born with DSD/intersex have never received a definite medical diagnosis and so may prefer to describe themselves using other terms (Jones et al., 2016). In this thesis, the DSD/intersex nomenclature will be used when talking about the participant group, the diagnostic names when necessary for clarification, and the term "affected individuals" when talking about people with DSD/intersex conditions. The term "disorder" will not be used in respect of people who feel that their congenital variation is precisely that—a variation—not something that needs to be pathologized into a disorder when others seek medical recognition (Jenkins & Short, 2017). An international professional network that promotes psychological well-being among people with variations in sex characteristics encourages people to "use terms that are specific to variation and may not identify with any umbrella term or overarching category" (PSIInternational, 2023). Moreover, the network states that "best practice is to ask about and use the language with which people understand themselves and to be mindful of the context in which one is speaking" (PSIInternational, 2023).

There is an ongoing debate as to which conditions should be covered by the umbrella term “DSD” (Lee et al., 2006). The 2006 consensus statement and later updates in 2016 suggests a DSD Classification dividing conditions in three groups: Sex Chromosome DSD, 46,XY DSD and 46,XX DSD (Lee et al., 2006; Lee et al., 2016), including hypospadias and sex chromosome DSD, such as Klinefelter and Turner syndromes. The definition used in the consensus statement in 2006 (Lee et al., 2006), as revised in 2016 (Lee et al., 2016), with that used in another consensus statement promoting holistic care (Cools et al., 2018) are integrated in this thesis.

DSD/Intersex: Historical Perspective

Prior to 1900, individuals with atypical sex development were little known (Dreger & Herndon, 2009). However, following medical advances and the development of gynecological science, there was increasing awareness that sex development could be diverse. Still, the external physical characteristics of the genitalia were regarded as the “true” sex of a person (Feder & Karkazis, 2008), and an individual with diverse sex development was expected to socially and sexually adhere to their assigned gender (Dreger & Herndon, 2009).

During the 1950s, hermaphrodites—that is, individuals who were born with atypical sex anatomy—became a subject of interest for surgery and medical treatments, and later many individuals with intersex variations underwent surgeries designed to assign them to either the male or female sex, often without their consent or understanding. (Colapinto, 2000; Feder & Karkazis, 2008). Later, three classificatory types of hermaphroditism were distinguished: male pseudohermaphroditism, which includes undervirilized 46,XY males; female pseudohermaphroditism, which includes overvirilized 46,XX females; and true hermaphroditism, which includes individuals with both ovarian and testicular tissue (Feder & Karkazis, 2008). The Johns Hopkins Hospital was the first hospital to offer a multidisciplinary approach toward DSD/intersex with the intention of treating DSD/intersex conditions in childhood so that there would not be medical or social problems in adolescence or adulthood (Lee et al., 2023).

It is important to emphasize that the definition of DSD/intersex variations used in the 1950s was heavily influenced by societal and cultural norms. There was a strong emphasis on assigning a clear gender identity to individuals, typically based on the dominant cultural understanding of gender. John Money, a psychologist working at the Johns Hopkins University, believed that gender was a product of nurture (upbringing and environmental factors) rather than nature (genes, chromosomes, and prenatal hormones). He claimed that as

long as a child was assigned a gender before the age of two, the child would confirm to that gender identity (Colapinto, 2000). As it is easier to surgically create female genitals than male, most undervirilized 46,XY male children underwent surgery to give their genitals a female appearance and assigned a female sex. However, history has shown Money's theory to be dubious at best, including the fact that contrary evidence was omitted from his work (Colapinto, 1997).

In the 1990s, activists sought to unite those who differed in terms of sex development and those who had similar experiences, which is why they started using the term "intersex," as introduced by feminist and biologist Anne Fausto-Sterling (Fausto-Sterling, 1993). The Intersex Society of North America (ISNA) defines a person as intersex "if she or he was born with a body that someone decided isn't typical male or female" but emphasizes that the definition depends on time, culture, gender, and the state of medical knowledge (Dreger & Herndon, 2009). For instance, prior to the scientific discovery of chromosomes, a female with XY chromosomes could not have been considered intersex. Many parents or caregivers (hereafter collectively referred to as parents), clinicians, and affected individuals did not recognize themselves, their children, or their patients in the "intersex" term, where sex is regarded as a continuum, something in between male and female, not as a two-sex system. During the 1990s, corrective surgery to make a DSD/intersex baby fit the gender assigned at birth was normal, while what had been considered ambiguous was in most cases corrected.

DSD/Intersex: Activist and Cultural Perspectives

DSD/intersex activists are individuals and groups who advocate for the recognition and rights of people with sex variations. They comprise individuals with DSD/intersex conditions, allies, and advocates, including medical professionals. They work to raise awareness of DSD/intersexuality, challenge harmful medical practices, promote bodily autonomy, and achieve societal acceptance and inclusion. Activists also work to challenge the binary understanding of gender, to promote awareness of the diversity of human bodies, and to campaign for informed consent, ensuring that people with DSD/intersex participate in their medical decisions (Interactadvocates, n.d; OIIeurope, 2023).

Culture can affect the lived experiences of individuals with DSD/intersex in several ways. Norwegian culture is known for its emphasis on egalitarianism, social justice, and protection of human rights, which can positively affect the experience of living with DSD/intersex conditions in the country. In the period from 2015–2019, attention was drawn to the unfulfilled rights of children with DSD/intersex conditions in Norway. This was

documented by a report on the challenges and opportunities concerning the law and DSD/intersex in Norway (Garland et al., 2018), a symposium on diversity in terms of sex development (Norwegian Directorate for Children, 2017), a report on the right of children to challenge gender norms in Norway (Egeland, 2016), and an article on the legal rights of an intersex child (Sandberg, 2016). These efforts seemed to raise little awareness as it fostered limited public debate, and DSD/intersex continued to be conditions mostly known to health-care professionals (HCPs) who work with affected individuals.

DSD/Intersex: Medical Perspectives

DSD/intersex are a group of conditions that involve variations in individuals' sex characteristics, causing their genitals, hormones, and/or chromosomes to differ from traditional conceptions of male and female bodies (Lee et al., 2006). Those who are born with a condition that affects sex development represent a diverse group of individuals with regard to their diagnoses, severity of medical complications, psychological impacts, treatments, and follow-up (Kim & Kim, 2012; Lee et al., 2006). DSD/intersex are complex conditions that affect not only physiological processes within the body but also feelings of identity and psychosocial well-being. Some patients have visible phenotype variations (e.g., in their genital appearance), while others exhibit genotype differences (e.g., in their sex chromosomes). Their conditions may have been discovered at birth or during childhood, adolescence, or adulthood, or they may even have gone undetected. Some patients are diagnosed due to severe illness as a child, others due to ambiguous genitalia, some due to delayed or missing menstruation or different development during puberty, and others again during the course of investigating the cause of difficulties in conceiving a child. The estimated incidence of DSD/intersex conditions with a genital appearance requiring genetic and endocrine investigation is 1:4500–1:5500 newborns (Sax, 2002); however, there are no clear estimates of how many people have a DSD/intersex condition in Norway, while figures from other countries vary among studies. Still, estimates indicate a prevalence of 0.5% worldwide (Lee et al., 2016). The related terminology lacks clarity because the phenotypes may be similar yet with different etiologies, making clinical classification problematic and, consequently, rendering the accurate estimation of incidence difficult (García-Acero et al., 2020). What we do know is that every year, approximately 10–20 newborns are referred to the two multi-regional treatment services in Norway due to questions about whether they may have a variation in sex development (Bjørndalen & Ræder, 2022).

Conditions Represented in the Thesis

In this thesis, individuals representing the three main DSD/intersex groups (see Cools et al., 2018) participated in the research (see Table 1). Each main group has several subgroups that have specific diagnoses e.g., Turner syndrome and MRKH. The medical aspects and the most typical features of the subgroup conditions that are represented in this study are briefly described in the following. Notably, the focus is on the most typical features of the conditions, meaning that what functions better in the lives of individuals with DSD/intersex conditions will receive less focus in the following descriptions.

Table 1. Examples of Classifications of Differences of Sex Development

Sex Chromosome DSD	46,XY DSD	46,XX DSD
A: 47,XXY (Klinefelter syndrome and variants)	**A: Disorders of gonadal (testicular) development** 1. Complete or partial gonadal dysgenesis (e.g., Swyer syndrome) 2. Ovotesticular DSD 3. Testis regression	**A: Disorders of gonadal (ovarian) development** 1. Gonadal dysgenesis 2. Ovotesticular DSD 3. Testicular DSD
B: 45,X (Turner syndrome and variants)	**B: Disorders in androgen synthesis or action** 1. Disorders of androgen synthesis 2. Disorders of androgen action (e.g., androgen insensitivity syndrome)	**B: Disorders of androgen excess** 1. Fetal (e.g., Congenital adrenal hypoplasia) 2. Fetoplacental 3. Maternal
C: 45,X/46,XY (mixed gonadal dysgenesis)	**C: Persistent Müllerian duct syndrome**	**C: Other/unclassified disorders** (e.g., Mayer-Rokitansky-Küster-Hauser syndrome type I and II)
D: 46,XX/46,XY (chimerism)	**D: Other/unclassified disorders** 1. Syndromic associations of male genital development (e.g., cloacal anomalies, hypospadias of unknown origin)	

Note: Classification of DSD proposed by Cools et al., (2018), Lee et al. (2006) and Lee et al. (2016).

Group 1: Sex Chromosome DSD

Individuals with sex chromosome DSD have a chromosome pattern that differs from the usual XY or XX. More specifically, they may have a missing X chromosome or an extra chromosome (XXY).

Klinefelter syndrome (description based on Skakkebæk et al. [2021]). Klinefelter syndrome (KS) has an estimated prevalence of 1 in every 660 males. The clinical features of KS vary widely, resulting in some patients being diagnosed prenatally, some in childhood, some in adulthood, and some remaining undiagnosed with few, non-specific, or no symptoms. Individuals with KS are born with an extra X chromosome (XXY) and do not produce the usual level of testosterone. Males with KS may therefore experience differences in the development of male characteristics (testes, muscle bulk, gynecomastia and body hair) and delayed puberty. KS may also affect bone strength and fertility. Although intelligence is usually unaffected, men with KF have an increased risk of anxiety, learning difficulties, and depression. Weak muscles, hypermobile joints, type 2 diabetes, and growing taller than expected are other features in males with KF. Some problems associated with KS can be treated with hormone replacement therapy (HRT) (testosterone gel, tablets, or injections), which can be started during puberty. Patients who have excess breast tissue are offered breast surgery reduction. While males with KS were previously considered infertile, intracytoplasmic sperm injection (where sperm are collected surgically and injected into the egg) has increased the chance of conceiving a baby with own genetic material by up to 25%. For others, donor semen or adoption can facilitate fatherhood.

Turner syndrome (description based on Gravholt et al. [2023]). A total of 1 in 2000–4000 phenotypic females are born with Turner syndrome (TS). TS occurs when one of the X chromosomes is missing, either entirely or partially. Even though the clinical features are heterogeneous and some may have milder symptoms than others, the most common features of TS are a short stature and non-functioning ovaries, resulting in delayed puberty, lack of monthly periods, and infertility. TS is also often associated with a number of other health conditions and symptoms, including learning difficulties and social problems, as well as with congenital malformations of the heart, skeletal structure, and kidney. TS can be diagnosed at every stage of life, although it is most commonly diagnosed during childhood, in the late pre-teen period (8–12 years), or in late adolescence/early adulthood. The median age at diagnosis is 15 years. Most females with TS require HRT, which serves to induce puberty, strengthen bone development, and promote the development of secondary and primary sex characteristics. HRT is provided via transdermal administration or tablets. Females with TS are offered growth hormone therapy to increase their height potential. While most women with TS cannot conceive on their own, successful pregnancies have been reported. Moreover, a woman with TS has the potential to conceive via oocyte donation (eggs from another woman) or using her own oocytes/eggs if, as a young woman/child, she froze eggs following

ovarian hyperstimulation. Females with TS require multidisciplinary care and treatment into adulthood, with a particular focus on the transition from childhood to adulthood.

Group 2: 46,XY DSD

Hypospadias. Hypospadias are common congenital malformations that affect the development of the urethra and penis (Giannantoni, 2011). The prevalence of hypospadias is around 1 in every 250 boys per year; however, the majority of cases are distal, whereas proximal hypospadias account for about 10–15% of cases (Baskin & Ebbers, 2006). The clinical features vary along a spectrum from the urethral opening appearing nearer the tip (distal), on the underside along the shaft, or nearer the scrotum (proximal). Moreover, the penis may be underdeveloped or have a stenotic opening and a chordee (a ventral curvature of the penis), with the latter being most commonly seen in proximal cases (Giannantoni, 2011). When there is a stenosis in the urethra, the urine flow can be weak. If required or wanted, the treatment for hypospadias is surgery. The goal of such surgery is to "create a functional penis adequate for sexual intercourse, produce a correct urethral reconstruction to allow the patient to stand to urinate, and offer satisfactory cosmetic results" (Giannantoni, 2011, p.1190). Surgery is usually performed within the first two years of life when the parents are proxy decision makers (van der Horst & de Wall, 2017). There are a variety of techniques available, although there is currently no "gold standard" surgical technique (Castagnetti & El-Ghoneimi, 2022; Diamond et al., 2017). The occurrence rates of complications such as fistulas, urinary tract infections, urethral strictures, curvature, stenosis, cosmetic issues, voiding dysfunction, and both sexual and psychosexual dysfunction vary among studies and with the age of the individual with hypospadias (Chen et al., 2022). Fertility is difficult to measure in this regard. The number of children is lower in populations of operated men with hypospadias compared to the general population. The reason for this is unclear and likely multifactorial as the semen quality of men with hypospadias is reported to be normal (Skarin Nordenvall et al., 2020).

Androgen insensitivity syndrome (AIS). AIS appears in two forms: complete androgen insensitivity syndrome (CAIS) and partial androgen insensitivity syndrome (PAIS). Only individuals with CAIS are represented in this study and will be described in this section. CAIS is very rare and has an estimated prevalence of between 1 in 20,000 and 1 in 99,000 individuals with a 46,XY karyotype (Oakes et al., 2008). Individuals with CAIS exhibit hormonal resistance to androgens, which results in a traditional female phenotype with a 46,XY karyotype and gonads that produce androgens (testosterone), although cells are resistant to the effect of testosterone. Consequently, individuals with CAIS have no uterus or

ovaries, and they have a shorter and blind-ended vagina. With the traditional female phenotype, most grow up as females, and the presentation of CAIS may be either primary amenorrhea in adolescence or inguinal hernia in childhood (Hughes et al., 2012). In terms of treatment, prophylactic removal of the gonads remains controversial, with the timing of the removal continuing to be debated, although the procedure is still considered due to the risk of developing gonadal malignancy (Tyutyusheva et al., 2021). HRT is mandatory after gonadectomy (Tyutyusheva et al., 2021). In today's clinical practice, vaginal dilatation is delayed until the individual seeks treatment for short vaginal length, and vaginal creation surgery is seldom necessary (Duranteau et al., 2021).

Swyer syndrome. Individuals with Swyer syndrome have a female phenotype with an unambiguous female genital appearance (King & Conway, 2014). They have a 46,XY karyotype with an uterus, vagina, and fallopian tubes. Swyer syndrome is estimated to affect 1 in 80,000 individuals with a female phenotype (King & Conway, 2014). The gonads have not developed as expected and produce no hormones. As a result, individuals with Swyer syndrome experience delayed puberty and amenorrhea, which often prompt medical investigation and lead to a diagnosis. Another clinical feature is increased height in adulthood (King & Conway, 2014). Individuals with Swyer syndrome cannot have genetic children, although pregnancy may be feasible via egg donation. Early prophylactic bilateral gonadectomies are discussed due to the risk of developing gonadal malignancy. Hormonal therapy with estrogen is required to induce puberty and the development of secondary sexual characteristics, such as breasts. Moreover, long-term estrogen and progesterone replacement therapy is offered (Meyer et al., 2019).

Group 3: 46,XX DSD

Congenital adrenal hypoplasia (CAH). CAH is an inherited condition affecting the adrenal glands that can be present in both 46,XX and 46,XY individuals (Nordenström et al., 2022). CAH is categorized as a DSD when the person with the condition has 46,XX chromosomes (Lee et al., 2006), which means that only 46,XX individuals are included in the present description. CAH is divided into two subgroups: classic and non-classic CAH (Witchel, 2017). The classic form occurs in between 1 in 10,000 and 1 in 20,000 newborns, while the non-classic form is more common (Auer et al., 2022). Furthermore, the classic form comprises two types: salt-wasting and non-salt-wasting CAH. Individuals who are born with CAH exhibit complete or partial insufficiency in terms of the production of an enzyme (most often 21-hydrooxylase) that is necessary for the body to produce cortisol and aldosterone, two

vital hormones. As a consequence of this enzyme deficiency, the body also overproduces certain androgens, such as testosterone. Symptoms of the associated hormonal imbalance are often evident within the first few days or weeks after birth, and babies can become very ill due to experiencing a salt-losing crisis (Nordenström et al., 2022). For individuals with XX chromosomes, the overproduction of androgens may result in virilization and genital variations, including a larger than typical clitoris, a urogenital sinus, and a closed vaginal opening (Lee et al., 2006). This may complicate the gender assignment process following birth. However, the symptoms will vary according to the degree of the enzyme insufficiency. Individuals with CAH require lifelong cortisol/glucocorticoid replacement therapy to stabilize their hormone levels, and some might find it challenging to achieve a balance between overtreatment and undertreatment (Nordenström et al., 2022). Medical management of genital variation has historically involved feminizing surgery, including creation of a vagina and separation of the urogenital sinus, a labiaplasty, and clitoral surgery (Shalaby et al., 2021). This treatment remains highly controversial and debated due to issues concerning informed consent, gender identity, and the risk of damage to adult sexual function caused by genital surgery, in addition to young girls not requiring a functional vagina (Auer et al., 2022). Still, while this treatment has become less routine, it is still performed in several Western countries (Shalaby et al., 2021). CAH is part of the newborn screening process in many countries (including in Norway since 2012), with the aim being to detect the condition shortly after birth and prevent severe illness or death due to an adrenal crisis (Tangeraas et al., 2020). Women with CAH have a lower birth rate when compared with the general population (Hirschberg et al., 2021). However, a multitude of factors may influence the ability and desire to become pregnant, including previous genital surgery, a non-heterosexual orientation, and psychosexual issues impacting sexual activity (Nordenström et al., 2022). In fact, a recent study indicated that reproductive outcomes are only impaired in women with salt-wasting CAH (Hirschberg et al., 2021).

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) (description based on Herlin et al. [2020]). MRKH has an estimated prevalence of 1 in 5000 females, and it has a female karyotype (46,XX). The ovaries and external genitals are the same as with traditionally female phenotypes. Moreover, puberty debut is the same as for peers, except for primary amenorrhea, as 46,XX individuals with MRKH have a uterus, cervix, and upper vagina that has not developed as expected. Consequently, they do not start to menstruate and cannot become pregnant. Other clinical features of MRKH include renal and skeleton malformations as well as uterine remnants where, in some cases, surgical removal is considered. Cardiac

abnormalities and hearing loss are reported in fewer than 5% of individuals with MRKH. Vaginal dilation using dilators or dilation via intercourse is recommended as a first-line treatment if necessary and desired to increase vaginal length, although vaginal creation surgery is available for patients who experience dilation therapy failure. Motherhood can be achieved, if wanted, by an individual with MRKH through adoption, surrogacy with oocytes, or in recent years, uterus transplantation. However, uterus transplantation is still experimental and not available in Norway.

Outcomes of DSD

The outcomes of DSD/intersex can encompass many domains of physical functioning, sexual health, and psychosocial well-being. Moreover, different outcomes can be the result of different etiologies, surgical approaches, medical treatments, and reactions of others, or triggered by the condition itself. A more complete explanation of the outcomes in types of domains are provided below for understanding the lived experiences of individuals with DSD/intersex.

Management

Some DSD/intersex conditions do not require any medical care except for the provision of thorough information, availability of psychosocial care, and potential to address any concerns that might arise. For others, medical care involves replacing missing hormones (sex hormones and/or corticosteroids), providing emotional and psychosocial support, and discussing potential surgical interventions. In Norway, the diagnosis, follow up, and treatment of children with DSD/intersex are organized by two multidisciplinary teams, which are located at the Oslo University Hospital and Haukeland University Hospital, up to the age of 18 years. After that age, no organized multidisciplinary follow up is available.

Medical treatment. For affected individuals and for the parents of young children who require HRT, mimicking a natural hormone level during an average day and during the entire life cycle is a complicated and both time- and energy-consuming activity (Nordenström et al., 2022). Lifelong hormonal treatment is indicated in relation to some conditions, for example, in individuals with CAH, while it is indicated between puberty and the time when menopause usually occurs to others. Furthermore, as DSD/intersex conditions are rare, complex, and heterogeneous, individuals often require individualized care and management by an experienced multidisciplinary team throughout their lives (Cools et al., 2018).

Surgical management. The surgical management of genitalia with a diversity in terms of the phenotype and gonads is controversial. Evidence both for and against this type of surgical treatment is conflicting, making it difficult to reach a clear consensus (Cools et al., 2018; Mouriquand et al., 2016). Decisions regarding so-called feminizing surgery have traditionally been influenced by the degree of virilization of the external genitalia, which can be graded according to the Prader staging, where 0 indicates typical female external genitalia without clitoromegaly and 5 denotes complete virilization (Auer et al., 2022). Feminizing surgery has traditionally been performed to provide the genitalia with a female appearance, to reduce parental stress, to reduce the risk of psychosocial stress in affected individuals, and to enable sexual intercourse in adulthood (Auer et al., 2022). Issues such as the child's right to provide informed consent, bodily integrity, the risk of short- and long-term complications (e.g., unsatisfactory clitoral sensation and sexual function), and the irreversibility of the surgery have all raised concerns (Auer et al., 2022; Claahsen-van der Grinten et al., 2022; Rapp et al., 2021). Even though surgical techniques have improved (Lee et al., 2016), studies of the long-term outcomes still indicate complications affecting sexual health in a negative way (Almasri et al., 2018; Nordenstrom et al., 2010; Rapp et al., 2021).

Surgery to correct undervirilized male genitals (proximal hypospadias) raises less concerns because no tissue is removed (Rapp et al., 2021), although there is still a high risk of complications (e.g., fistulas of the urethra, meatal stenosis and strictures over time). Indeed, a large multicenter quantitative study reported complication rates close to 50%, including loss of glans sensitivity in about 5% in what is considered severe cases (Long & Canning, 2016). While advances in surgical techniques and anesthesia have led to reduced complication rates following proximal hypospadias repair, including lower rates of urethral strictures and fistulas (Snodgrass et al., 2014), there remains a risk of such complications (Chen et al., 2022), and some patients will require multiple reoperations (Andersson et al., 2020). An additional challenge stems from the fact that certain complications manifest during the later stages of adulthood, often going unnoticed or untreated, particularly when patients find themselves navigating the transition from pediatric to adult urology services (Chen et al., 2022).

While the consensus statement from 2006 (Lee et al., 2006) and the update from 2016 (Lee et al., 2016) advise reducing the surgical management of external genitals, there is little evidence in support of a considerable change of approach in recent years (Cools et al., 2018; Creighton et al., 2014; Mouriquand et al., 2016; Wolffenbuttel & Crouch, 2014). However, some countries, such as Iceland and Malta, have introduced a ban on non-consensual and non-therapeutic interventions that are not considered vital for the health of children born with

DSD/intersex (not hypospadias) until they are old enough to give informed consent (Alaattinoglu, 2022). In 2016, the United Nations issued a call for "Governments to prohibit forced and coercive surgeries and other medically unnecessary treatments on intersex children without their consent" (Office of the High Commissioner for Human Rights, 2019). Cools et al. (2018, p.421) stated that attitudes towards reconstructive surgery has "changed dramatically following disquieting reports of unfavorable outcomes, including high complication and/or reoperation rates and patient dissatisfaction" Despite this, reports have contested whether practice have changed (Michala et al., 2014).

Evidence for or against gonadectomy is contradictory, meaning that the surgery remains a subject of debate due to uncertainty regarding its benefits and risks, its irreversible nature, and the lack of consensus (Cools et al., 2018; Lee et al., 2016). Gonadectomy is performed when it can mitigate the risk of gonadal tumors, reduce gender-contrary hormonal production, and provide psychosocial benefits by aligning the patient's external appearance with their gender identity (Lee et al., 2016; Wisniewski et al., 2000). Conversely, critics have noted its potential negative effects on fertility and hormonal balance, emphasizing the importance of shared decision making and careful consideration of individual cases (Cools et al., 2018; Hughes et al., 2006). The risk of malignancy is reported to be very low in some DSD/intersex conditions, and the gonads should therefore be left in place (Cools et al., 2018). In other conditions, there is more uncertainty regarding the risk of malignancy and "there are no useful markers to predict the tumor risk other than gonadal histology" (Duranteau et al., 2021, p.173). However, some conditions demonstrate a high malignancy risk, and gonadectomy is recommended (King & Conway, 2014) Recent studies have underscored the need for a personalized approach to gonadectomy in DSD/intersex, balancing medical concerns with patient autonomy and long-term well-being (Kyriakou et al., 2016; Rapp et al., 2021).

DSD/Intersex: Psychosocial and Psychosexual Perspectives

In both the original consensus statement from 2006 and the revised version from 2016, the importance of a psychosocial focus of living with DSD/intersex is highlighted (Lee et al., 2006; Lee et al., 2016). Earlier research on DSD/intersex was dominated by a medical focus, while psychosocial research has traditionally focused on brain organization theories (Liao & Roen, 2014; Roen & Pasterski, 2014). Furthermore, the data on quality of life (QoL), which is often embedded in quantitative methods, and psychosocial outcomes in those born with a DSD/intersex condition vary widely, depending on the condition, size of the studied cohort,

diagnostic criteria used, and studied time period, as well as on the potential cultural aspects and their influence on reactions to the diagnosis. Some studies reported equal or better QoL than the general population (Rapp et al., 2018), whereas others showed poorer QoL (De Vries et al., 2019; Engberg et al., 2015; Waehre et al., 2022). However, individuals' experiences may be unified by a lack of information, difficulties with disclosure, sexual health issues, and stigma, which can affect individuals' psychosocial health and QoL (Hughes et al., 2006; Hughes et al., 2007; Lampalzer et al., 2021; Malmqvist & Zeiler, 2010; McCauley, 2017; Weidler & Peterson, 2019; Wisniewski et al., 2019). In fact, Waehre et al. (2022) found significantly lower levels of psychological distress in adults with DSD/intersex conditions who had positive experiences with information given prior to surgery than in adults who had mainly negative experiences.

A positive body image and good self-esteem are associated with greater disclosure in individuals with DSD/intersex conditions (van de Grift et al., 2018). However, disclosure may also be associated with ambivalent feelings (Lampalzer et al., 2021; Sharratt et al., 2020), while the expectation of being stigmatized may have an influence on disclosure too (Ernst et al., 2016).

Sexual health was reported to be satisfactory in some quantitative studies (Engberg et al., 2022; Schonbucher et al., 2008), while other studies, both quantitative and qualitative investigations, reported the negative impacts of the medical consequences of DSD/intersex (type and severity), the psychological experiences of treatment (e.g., distress, anxiety), and the reactions of others (e.g., stigma) (Liao et al., 2011; Meyer-Bahlburg et al., 2018; Sani et al., 2019; van de Grift et al., 2022).

A qualitative approach to understanding personal experiences of DSD/intersex conditions is becoming more visible in the research literature, as is activists' and human right defenders' approach to letting affected individuals of all ages be heard (e.g., Denver, 2004; Hart & Shakespeare-Finch, 2022; Suorsa-Johnson et al., 2022; Zeiler & Wickström, 2009). In this regard, Hart and Shakespeare-Finch (2022) recently described the experienced trauma of DSD/intersex and post-traumatic growth in individuals with DSD/intersex conditions, revealing how lacking both everyday language and peers to share experiences with complicate the healing process. These findings confirmed those of Lundberg et al. (2016), who highlighted an important factor regarding successful information transmission to be the information provided being perceived as relevant and experienced as meaningful, in addition to affected individuals being able to explain it to others. The feeling of being stigmatized may also be a result of experiences during childhood and adolescence in health-care settings

(Meyer-Bahlburg et al., 2017). Stigma and disclosure may be closely linked, and they have the potential to influence the lived experiences of individuals. Ernst et al. (2016) found that fear of rejection or being labeled a “freak” (expected stigma) was a common barrier to sharing diagnosis-related information among individuals with MRKH.

Theoretical Perspective

This chapter provides a theoretical framework with which the present research can be understood. As each paper included in this thesis provides information about relevant research within its specific frame of reference, the purpose of this introductory section is to present a comprehensive overview of the framework underpinning the understanding of living with a DSD/intersex condition, thereby creating a coherent thread linking the various other sections. In addition, a reflection on how the concepts of stigma, epistemic injustice, and healing have influenced this work will be provided.

Lived experiences refer to the way in which something presents itself or is experienced by an individual (van Manen, 2017). According to van Manen (2017), lived experiences refers to the "raw" experiences or phenomena we seek to understand. The intention when exploring lived experiences is to move beyond the taken-for-granted understanding of a given experience or phenomenon, in this case of individuals with DSD/intersex conditions.

Stigma and Living with DSD/Intersex Conditions

The concept of stigma is prominent within the realm of chronic conditions (Engebretson, 2013). Stigma has the potential to contribute to poorer psychological health. As a result, living with a chronic condition and expecting stigma make affected individuals less likely to access health care (Earnshaw & Quinn, 2012), which may lead to an decrease in their adherence to therapy (Eisenberg et al., 2009). As defined by Goffman (1963) and further expounded upon by Link and Phelan (2006), stigma entails the assignment of both stereotypical traits and undesirable attributes to certain groups, which leads to social differentiation when other people seek to avoid such traits and attributes. This is a result of a social process whereby we attach certain attributes to groups of people, individuals, or behaviors (Joachim & Acorn, 2000), which creates a distinction between us and them (Link & Phelan, 2006). Goffman (1963) described stigma in terms of three perspectives: internalized stigma, which entails the application of negative beliefs and stigmatic feelings to oneself; anticipated stigma, which concerns the expectation of being a target of a stereotype, prejudice, or discrimination; and experienced stigma, which involves day-to-day experiences of stereotyping, prejudice, and discrimination from others (Earnshaw & Quinn, 2012; Fox et al., 2018). Structural stigma, a concept used to describe the societal norms, laws, and policies underlying stigma that restrict the opportunities and resources of stigmatized groups or fail to protect their rights, has frequently been used in the transgender literature and personal

testimonies (e.g., Falck & Bränström, 2023; Human Rights Watch, 2017). Structural stigma is said to also be relevant in relation to DSD/intersex groups, for example, misconceptions and generalizations concerning the involvement of individuals with DSD/intersex conditions in sport or the belief that DSD/intersex is a gender identity (Karkazis & Carpenter, 2018; Martínez-Patiño, 2005). Structural stigmas may increase other stigma conceptions, and they may represent an indicator of adverse health outcomes among stigmatized individuals (Falck & Bränström, 2023). For instance, individuals may read about athletes with CAIS who have been banned from championships and who are described as non-women in the media. A 16-year-old individual who identifies as a woman and who has been diagnosed with CAIS may not want to be associated with such descriptions and therefore may avoid telling others about her condition, which could reduce her self-esteem.

Some 60 years since Goffman's (1963) pioneering publication, "Stigma: Notes on the management of spoiled identities," the theory remains influential and relevant in the DSD/intersex community. Recent studies have examined the effects of stigma on people with DSD/intersex conditions and identified both feelings of shame (i.e., experienced stigma) (Engberg et al., 2016) and withdrawal behavior (i.e., anticipated stigma) to limit people's ability to integrate with others (Meyer-Bahlburg et al., 2018; Meyer-Bahlburg et al., 2017; van de Grift, 2023). Most individuals who are born with DSD/intersex conditions can "pass as normal" because their conditions are "hidden" or invisible with the naked eye. However, the stigma remains real as long as there is the potential for disclosure (Goffman, 1963). Thus, issues related to disclosure and stigma may be visible in social contexts, and they may result in communication difficulties because stigma occurs in social interactions (Bos et al., 2013; Earnshaw & Quinn, 2012). An extra burden of concealable conditions is the fact that individuals often live with the fear that other people will find out and subsequently misunderstand, discriminate against, or bully them (Engebretson, 2013; van de Grift, 2023). Society tends to generalize from a particular condition to other attributes (Joachim & Acorn, 2000). For example, when people who are unfamiliar with DSD/intersex believe that those born with DSD/intersex conditions must identify as transgender or consider themselves to have been "born in the wrong body" (Hegarty et al., 2021).

Affected individuals and/or their families may expect stigmatization, either because of previous experiences of stigmatization or because of an internalized process of stigma (Quinn & Chaudoir, 2015). As their conditions are concealable, they might have heard or experienced unfavorable or ignorant reactions to other people who challenge gender norms (due to bodily variations that lie outside the binary ideals of male and female). For example, if there were

uncertainties about their gender at birth due to under virilized genitalia and they hear others talk about those previously called "hermaphrodites" in a ridiculous way. They may even exhibit similar reactions and stereotypes in relation to others, not knowing that they are "one of them" and only later discovering that their female sex was not what they thought due to them being born with XY chromosomes.

Concealable stigmatized conditions are likely to increase psychological distress and, consequently, to influence how individuals react to and cope with stigma (Quinn & Chaudoir, 2015). Those who have an invisible or concealable stigmatized attribute may have less chance to receive peer support from others with the same condition/experience, meaning that they may miss the opportunity to enjoy the social and psychological support that peers can provide (ibid). Moreover, a person with a concealable difference that they have not disclosed must decide how to handle information that could possibly lead to stigmatization (Joachim & Acorn, 2000). Quinn and Chaudoir (2015, p.5) described how the effect of stigma on an individual's psychological well-being varies in terms of "whether the identity is considered to be critical to their self-definition." An individual who identifies as a woman based on the ability to give birth may have greater difficulty coping with a DSD/intersex condition than an individual whose identity centers on what they can accomplish as a friend or an athlete. Quinn and Chaudoir (2015, p.5) referred to this as "centrality"—that is, "how central the identity is to the self". Power differences may also affect the feeling and production of stigma (Parker & Aggleton, 2003). Such power differences will be explored later in this thesis (the chapter "Epistemic injustice").

Body Image

The body is both something "we have" and something "we are" (Leder, 2022). The importance of taking good care of your body and presenting it in a positive light as well as the fact that bodily diversity is talked about more openly in Western cultures reflect notions of how the body is both an object (something we have) and a subjective lived experience (what we are). In this context, body image refers to the perceptions, thoughts, and feelings an individual has regarding their own physical appearance (Kling et al., 2019). Body image and stigma are interconnected, as both societal norms and beauty standards often contribute to the stigmatization of individuals who do not fit the mold. This can lead to a person having negative perceptions of himself, which may potentially cause mental health issues.

Living with a DSD/intersex condition can be challenging when an individual's body diverges from the current ideals of being healthy, attractive, "normal," and fitting into the

dichotomy of the two-gender model (Human Rights Watch, 2017). Body satisfaction and appearance represent important elements of how individuals perceive themselves, meaning that they are major contributing factors to self-esteem (Tiggemann, 2011). Body image is closely related to self-esteem (Kling et al., 2019), and both a positive body image and good self-esteem are important for an individual's QoL (Tschaidse et al., 2022). In recent years, studies have increasingly focused on understanding the complex relationship between body image and DSD/intersex. In this regard, different factors have been found to affect the body image of people with DSD/intersex conditions, including the degree of virilization in females (Callens et al., 2021), body mass index and increased waist–hip ratios (Falhammar et al., 2007), limited disclosure (van de Grift et al., 2018), genital appearance (Schonbucher et al., 2008), and degree of virilization at birth (Kanhere et al., 2015; Krege et al., 2022; Warne et al., 2005). In addition, Wisniewski et al. (2019) emphasized the need for health-care providers to address body image concerns early on following a DSD/intersex diagnosis, with the aim being to improve patients' psychological well-being. However, a cross-sectional study noted a fairly positive body image among a large heterogenic group of individuals with DSD, although it was still lower than the control values (van de Grift et al., 2018). Another study, which investigated physical satisfaction with different body parts (Body Image Scale) among women with DSD, observed greater dissatisfaction with their bodies than among the women in the matched control group (Ediati et al., 2015). In the same study, both men and women were dissatisfied with their genital appearance and with other sex-related body parts (Ediati et al., 2015). Body image is said to have a greater impact on the self-esteem of women when compared with men (Tiggemann & McCourt, 2013). Again, low self-esteem and low body image satisfaction may influence or increase an individual's feeling of being a stigmatized (Zhang et al., 2020).

Epistemic Injustice

When parents learn of their child's DSD/intersex condition, or when those born with a DSD/intersex condition are told of their diagnosis, such information may change taken-for-granted notions of sex and gender. For some, it might engender a feeling of not knowing who to talk to or what to talk about regarding the condition (Gough et al., 2008). According to Gough et al. (2008), the parental experience of having a child with DSD/intersex condition can be described with the phrase: "They (parents) did not have a word." The concept of epistemic injustice provides a lens through which to view these experiences, where both knowledge and power differentials impact communication. Epistemic injustice describes how

someone with knowledge or power has an advantage over someone who does not have such knowledge or power (Carel & Kidd, 2014). Fricker (2017) identified two forms of epistemic injustice: hermeneutical and testimonial. *Hermeneutic injustice* occurs when a person cannot make sense of their own lived experience because of this gap in knowledge (Carpenter & Jordens, 2022). An example of hermeneutic injustice relevant to the present work is that parents and individuals with DSD/intersex conditions often lack sufficient understanding of the DSD and intersex concepts (Feragen et al., 2019). This can lead to affected individuals and their families missing out on peer support from the wider DSD/intersex community due to only searching for information about their specific condition. In addition, Carpenter and Jordens (2022) addressed the question of whether someone can provide informed consent to DSD/intersex-related surgery without having access to information about, for example, CAH being defined as a DSD/intersex condition. *Testimonial injustice* occurs when prejudice hinders someone from giving justice, meaning, or credibility to a speaker, phenomenon, or situation due to interpreting its meaning in accordance with their prejudice (Fricker, 2017). This can result in the listener assigning either too little or too much credibility to the speaker (Carel & Kidd, 2014).

Fricker's (2017) concept of epistemic injustice can be applied to the lived experience of those with DSD/intersex conditions and to elucidating the phenomena of disclosure, communication with others, understanding and knowledge dissemination, and shared decision making. An example of epistemic injustice can be seen in the experience of South African 800 m Olympic champion Caster Semenya, who was prohibited from competing by new rules of the International Association of Athletics Federations (IAAF). This matter was written about in newspapers (e.g., in *Aftenposten* [Saugestad, 2022], where women with XY DSD were described as biological men) and sparked a debate as to whether women with DSD/intersex conditions and transgender women should be allowed to compete in women's sports. Sebastian Coe, the IAAF president, stated that "The core value for the IAAF is the empowerment of girls and women through athletics" (Goh et al., 2023). Still, women with naturally high androgen levels have to take medication to lower their blood testosterone levels if they are to be eligible to compete. This simplification of what defines a woman (or a man) represents an epistemic oversimplification. I will not use the concept of epistemic injustice to, for example, discuss the pros and cons of disclosing information about a DSD/intersex conditions, or the pros and cons of DSD/intersex-related surgeries, although I will use it to discuss how this perspective can help us understand the lived experiences of people with DSD/intersex conditions.

Navigating Stigma Through Healing

Phenomenological perspectives on the lived experiences of chronic conditions often highlight the subjective experience of how it feels to be ill (illness) rather than presenting a disease or sickness perspective (Eisenberg, 1977). Disease is often identified by detecting "abnormalities in the function and/or structure of body organs and systems" (Eisenberg, 1977, p.9). Illness focuses on the individual's lived experiences as accessed via personal accounts, disease is understood as something that can be identified through "biological studies and analyses of the physiological state of our bodies" (Svenaeus, 2005, p. 28). Illness cannot be defined by an objective truth, whereas disease can be examined, predicted, and measured (Hofmann, 2016, p. 17). Sickness represents a third dimension of understanding lived experiences. It is connected to language and cultural aspects, and the understanding of sickness is often shared by a social group (Hofmann, 2016, p. 17). Notably, a person can experience illness without being diagnosed with a disease, while a disease can occur without a person feeling ill (Eisenberg, 1977). Examples on the latter include when a person is diagnosed with PAIS due to accidental findings on an ultrasound/chromosome test, or when a baby is diagnosed with hypospadias due to the altered appearance of the penis/urethra but no signs of illness.

Living with a DSD/intersex condition, which will inherently be a chronic and rare condition, involves coping with that condition as well as with the reactions of other people who are unfamiliar with the condition. These unfamiliar others may include family, friends, coworkers/students, and HCPs. Prior studies have found that both visible and concealable differences can be stressors for the individual, and they can result in the use of coping mechanisms (Chaudoir et al., 2013). Here, concealable differences are more likely to lead to the use of avoidance strategies (Hatzenbuehler et al., 2009).

When viewed through the lens of phenomenological theory, living with a DSD/intersex condition becomes a process whereby the diverse body shifts in and out of focus based on social interactions with others (Leder, 1990). The medical doctor and philosopher Drew Leder (2022) can contribute to understanding lived experiences of the importance of being normal—that is, a process of coping or healing when the diverse body remains or shifts in our consciousness. The coping strategies described in the literature often rely on clinical protocols concerning adaptive strategies for coping (White et al., 2018). Leder (2022) sought to demonstrate that even though having more coping strategies might mean you are better equipped to face challenges, each coping strategy might have a downside if used inappropriately, which has received less attention in the psychology field. In addition, the way

diversity is interpreted affects which "healing strategies become available to help the sufferer cope with long-term challenges" (Leder, 2022, p. 141), whether conscious or unconsciously. Leder (2022) applied a phenomenological analysis to clinical practice and divided the available healing strategies into those "freeing oneself from the body" and those that "embrace the body," where each strategy has its own advantage and disadvantages. Healing indicates building up dimensions of your life that represent losses and that have been damaged by illness or disease, whereas coping indicates adapting to the presence of illness or disease (Leder, 2022, p.19).

When the body experiences illness, it can be perceived as an unhomelike feeling (Svenaeus, 2005). Leder (2022) emphasized that dealing with this kind of "homelessness" requires different healing strategies. Individuals with a variety of DSD/intersex conditions may find the same strategies helpful, while individuals with the same condition may find different strategies useful. In the following subsections, two strategies for healing—namely, freeing oneself from the body and embracing the body—based on the relationship between the "self" and the "body" will be described. Freeing oneself from the body implies that the body is something *I have,* something I try to remove myself from. By contrast, embracing the body implies to moving toward and befriending the body, it is something *I am*, even though it gives me illness.

Removing Oneself from the Body

Ignoring a diverse body is one strategy people may use when their body causes them problems (Leder, 2022). Using the concept of ignoring, Leder (2022) explained how we tend to focus more on strength, which may shift into our consciousness, rather than on what causes problems. This may be a strategy for destigmatizing the body (Goffman, 1963) by focusing on positive aspects and ignoring illness, such as a concealable difference. What we focus on tends to occupy our consciousness and grow in strength. Even though ignoring something might seem like a simple way to make it go away, it can also prove valuable if, for instance, focusing on positive feedback from others helps you overcome your fear of intimacy. In Leder's (2022, p.141) words, it is "one of the most valuable healing strategies" as the problem recedes. Health-care personnel may increase the relevance of this strategy by focusing on treatment options rather than on the patient can accept their body.

This corresponds with the findings of Guntram's (2013) study of how women with two DSD conditions emphasize that they are just like any other women. They normalize their conditions' impact on their experience of feeling like a woman with reference to the

possibilities of making their body more like other women's bodies with the help of medicine and medical interventions, including surgery. Conditions such as DSD/intersex can be viewed as a biological dysfunction of a body part or system (Boorse, 1997, p. 4), when measuring the severity of a condition in relation to static parameters such as blood levels or penis/vagina length, we risk comparing each body to the statistically "normal" body (Banerjee, 2011). This may lead to adolescents and young adults (AYA) feeling dissatisfied with their body image and sexual health, which could compromise their QoL and psychological well-being. Individuals with a concealable condition such as DSD/intersex may experience being devalued and stigmatized when measured against what is considered "normal," and this may lead them to anticipate stigma if their condition becomes known (Quinn & Chaudoir, 2015), which may result in further concealment.

Another way of healing described by Leder (2022) involves *objectifying* the ill body. This strategy is influenced by a biomedical perspective and a disease-based approach to living with a condition, which offers individuals a sense of distance and control (Leder, 2022). The cause of a person's symptoms is sought, and a biomedical view of the body helps find cures for those symptoms (Leder, 2022). The body is something "I have," something I seek to fix. Objectification can be a strategy for distancing oneself from the diseased body by objectively understanding what is causing the diverse body to be something "I have" rather than something "I am." However, it can increase a person's distress to interpret every discomforting change in their body as a disease. Furthermore, when you cannot change what feels abnormal in a body, such as a missing uterus, your distress might increase. Zeeman and Aranda (2020) presented a review of intersex health and health-care inequalities, problematizing how the use of the diagnostic label "disorders of sex development," as classified in the World Health Organization's International Classification of Diseases (ICD-11), contributes to medicalizing what some people with DSD/intersex conditions define as natural variations. Moreover, other studies demonstrated that affected individuals and their parents prefer to use the name of the condition (e.g., CAH) due to fear of stigmatization (Feragen et al., 2019).

Embracing/Befriending the Body

When experiencing disease or illness, a person might lose their sense of control over and contact with their body. One way to reconnect with the body, which is the opposite of seeking distance from the body, is to embrace it. Leder (2022) described the related healing strategies as means of embracing the ill body: *accepting* what cannot be changed, *listening* to

the unconscious signs the body is sending, *befriending* the body by actively demonstrating an attitude of care, and *witnessing* attempts to understand suffering and where it comes from, not only its biological origins but also how memories can enhance or decrease both pain and suffering (Leder, 2022). Importantly, Leder's (2022) description of the different healing strategies shows that they can be blended in a sophisticated way to fit different situations, different individual personality traits, and different times of use.

This chapter focused on two of the available strategies—namely, ignoring and objectification—while recognizing the interdependency of the others. This approach was chosen as the starting point for how healing when living with a DSD/intersex condition can be understood, which represents an understudied perspective.

Study Perspective and Research Aims

The overall aim of the work described in this thesis was to explore the lived experiences of individuals with DSD/intersex conditions as well as the dilemmas faced by HCPs who work with individuals with such conditions. To achieve this aim, the following research questions were addressed:

1. How do adults with DSD/intersex conditions experience everyday challenges and how do issues such as disclosure, information sharing, and stigma affect their daily life?
2. How do AYAs with DSD/intersex conditions experience intimacy and sexual health?
3. What are HCPs' perspectives on decision making regarding DSD/intersex-related surgeries?

Methodology

This Ph.D. research project sought to provide individuals who are living or working with DSD/intersex conditions an opportunity to tell their stories. To achieve this, we applied a qualitative research design and conducted in-depth interviews with both AYAs and adults with DSD/intersex conditions (Malterud, 2001) as well as focus group interviews (FGIs) with health-care professionals who are familiar with DSD/intersex (Hydén & Bülow, 2003). FGIs are useful for achieving a deeper understanding of participants' experiences, attitudes, and views, which can provide access to their thoughts and perceptions on the subject of interest through dialogue and interaction, allowing the creation of data within the group (Kitzinger, 1995).

The present Ph.D. project comprises three qualitative studies. Study I, involved qualitative in-depth interviews with 15 adults over the age of 30 years who were already participating in the Bufdir project. Study II, entailed a qualitative in-depth study of 11 newly recruited AYAs with DSD/intersex conditions. Study III, involved three FGIs with 14 HCPs who were familiar with DSD/intersex conditions.

This chapter outlines the ontological and epistemological approaches which have overseen the research design. Furthermore, it provides a description of the methods and data analysis applied in this research project, the lessons learned, thoughts on reflexivity, an elaboration of the ethical considerations, and methodological reflections.

Interpretive Phenomenology

As we sought to understand the meaning in the participants' world from a first-person perspective, this study was conducted in accordance with the perspectives of the interpretive phenomenological tradition, drawing on the descriptions offered by Kvale and Brinkmann (2009, p. 30). In medical research, the interpretive phenomenological tradition aims to explore and understand the lived experiences of individuals with respect to their health and illness, as based on a lifeworld perspective. The lifeworld is the world of lived experiences (Dahlberg & Dahlberg, 2020). This tradition is rooted in the broader field of phenomenology, which emphasizes the study of human consciousness. Of central importance is the concept of meaning in terms of how the participants describe their experiences as well as the mutual meaning assigned to experiences related to a certain phenomenon (van Manen, 2017). We do not attempt to develop explanatory models for the participants' perspectives; rather, we seek to understand their experiences as generated through the data analysis process and to achieve

a deeper understanding of the subject matter (Willig, 2013, p. 16). Moreover, we aim to find meaning in something that is not evident as well as to gain insight into the ways individuals make sense of their experiences, the meaning they attribute to those experiences, and the impact of such meaning on their actions, decisions, and coping strategies. This requires a latent interpretation of the text. For this, we rely on our skill as researchers, which is inevitably sculpted by our social position and theoretical lenses, to perform the interpretation. The focus of most prior studies has been on describing issues related to DSD/intersex-related treatment, patients' QoL, and the problematic nature of attempts to reach a consensus due to the diversity of DSD/intersex conditions etiology, the rarity of the conditions, and the lack of long-term psychological and medical follow-up data (Baratz & Feder, 2015; Lee et al., 2016; Machado et al., 2016). In this research, a multidisciplinary stance was adopted with regard to the participants' experiences of DSD/intersex conditions as they are lived. This stance allowed us to apply the attitude or orientation of phenomenology to approach the research subject from our multidisciplinary background as researchers.

To gain access to the lived experiences of individuals with DSD/intersex conditions, we needed to provide lifeworld material for the phenomenological inquiry, which could have been collected via interviews, observations, or written sources such as blogs, literature, or art (van Manen, 2016). We chose to draw on two types of interviews: individual interviews with affected individuals and FGIs with HCPs. These sources provided thick descriptions of the lived experiences of DSD/intersex conditions and of the dilemmas faced by HCPs who work with DSD/intersex conditions. Human experiences are more complex than what we were able to capture through interviews alone (van Manen, 2016). Nevertheless, we sought to explore taken-for-granted or hidden experiences, and an interpretative phenomenological approach guided us throughout this process. We aimed to uncover meaning rather than to gather facts (van Manen, 2016). Finding and interpreting meaning within material require both time and wondering (Braun & Clarke, 2021). This wondering lead us to not only generate themes sheared by the participants but also to discover non-obvious meaning that we did not necessarily anticipate at the start of the analysis process, as described by Braun & Clarke (2021).

Research Team

To achieve the aim of the present study, we required the knowledge of, and collaboration with, several groups and individuals with varied professional expertise and different lived experiences with regard to DSD/intersex conditions.

The **researchers** comprised the four authors of all the published papers (SS, LF, AW, and LM) and a project manager from the Center for Rare Disorders (KBF), who co-authored Papers I and III. In addition, a colleague with a master's degree in psychology (CH) co-authored Paper I. All the researchers have extensive research experience, different clinical experiences, and experience in qualitative research. The researchers participated to different extents in planning, conducting, analyzing, and writing the research.

Two different **reference groups** were included to ensure patient and public involvement. In Study I, the reference group comprised representatives of patient organizations, organizations for lesbian, gay, bisexual, transgender, and intersex (LGBTI) people, and professionals with legal, medical, and psychological backgrounds (Table 2). In Studies II and III, we chose to include individuals with personal experiences of living with DSD/intersex conditions. Three AYAs and one adult, who were born with CAH, MRKH, hypospadias, and TS, respectively, participated. Among the four, three identified as female and one as a male.

The interview guide, nomenclature, and recruitment strategy were discussed in the initial meetings, while the analysis, results, and report writing were discussed in the later meetings. During Studies II and III, we also included the reference group in discussions regarding how and to whom to convey the knowledge acquired through the research project.

Table 2

Reference Group in Study I

Reference Group, Study I: Organizations/Representatives	**Participated**
Norwegian Directorate for Children, Youth and Family Affairs	X
Norwegian Association for Turner Syndrome	X
Harry Benjamin Resource Center	X
Fri (Norwegian Organization for Sexual and Gender Diversity)	X
Skeiv verden (Queer World)	X
Skeiv ungdom (Queer Youth)	X
Frambu (National Competence Center for Rare Disorders)	X
Oslo University Hospital (Child Endocrinologist)	X
University of Oslo, Faculty of Law	X
University of Oslo, Faculty of Psychology	X

Recruitment and Participants

In **Study I**, individuals who had already been recruited in 2018 and interviewed for the Bufdir project were recruited. This recruitment was performed with the help of clinicians at the Oslo University Hospital (OUS) and the Haukeland University Hospital (HUS), the two multidisciplinary DSD teams in Norway. All eligible participants were identified by searching for relevant diagnostic ICD-10 codes within their institutions' electronic health records. Individuals with 46,XY DSD and 46,XX DSD, which are registered at the Centre for Rare Disorders, and individuals with TS and KS, as registered with the Frambu Competence Center, were also invited to participate. Some conditions have patient support groups (TS, KS, MRKH, and CAH), which assisted with recruiting participants through their patient registries and/or distributing information about the study. In addition, organizations for LGBTI people promoted participation by spreading information about the study on their websites and social media channels. The recruitment was performed from April 2018 to August 2018, and the interviews were conducted from May 2018 to September 2018. The inclusion criteria for the Bufdir project were that participants had experiences of living with a DSD/intersex condition and were able to speak Norwegian. A total of 334 invitations were distributed, and 83 signed consent forms were received (aged 18–70). The response rate was higher among some groups (TS and KS) than others. A purposive sample of 27 people was chosen to ensure the participants were representative of different ages, genders, geographical affiliations, and diagnoses. Among these 27, 15 people were included in Study I.

The participants in Study I comprised 10 females and 5 males, all of whom reported identifying with the sex they were assigned at birth (either male or female). None identified as non-binary. The participants ranged in age from 30 to over 70 years (Mean age = 44 years). The participants came from different parts of Norway. Six participants had been diagnosed in infancy, four in childhood, two as adolescents, and three as adults. They were diagnosed with five different DSD conditions, representing all three DSD subgroups (46,XY DSD, 46,XX DSD, and sex chromosome DSD). Of the 15 adult participants, seven reported having had DSD-related surgery to the inner genitalia, outer genitalia, and/or gonads.

For **Study II**, AYAs aged 16–26 years were recruited from two subgroups of DSD: 46,XY DSD and 46,XX DSD. Clinicians working within the multidisciplinary DSD/intersex teams at the two hospitals (OUS and HUS) identified all eligible participants by searching for relevant diagnostic ICD-10 codes within their institutions' electronic health records. In addition, AYAs who had an appointment at an outpatient clinic at either of the hospitals during the recruitment phase also received an invitation to participate in the study. The

recruitment period ran from August 2020 to June 2021. In this study, individuals who had ongoing or previous contact with medical practitioners in the DSD/intersex teams were recruited. This was due to our aim of understanding AYAs' experiences of the multidisciplinary DSD/intersex teams, aid, and support, as well as their understanding of sexual health and surgical interventions.

Thirteen AYAs consented to participate. Among them, one had a sex chromosome DSD and was excluded. Another participant did not attend the scheduled interview. The recruitment might have been affected by the COVID-19 restrictions. Several of the outpatient consultations were postponed due to the pandemic, resulting in fewer AYAs receiving invitations to participate, or the AYAs might have cancelled the clinical follow ups themselves. The final 11 participating AYAs comprised eight females and three males, all of whom reported identifying with the sex they were assigned at birth (either male or female). None identified as non-binary. The participants ranged in age from 16 to 26 years (Mean age = 20.3 years). The participants came from different parts of Norway. Six participants had been diagnosed in infancy, three in childhood, and two as adolescents. None of the AYAs had received a diagnosis in adulthood. They were diagnosed with five different DSD conditions. Of the 11 AYA participants living with a DSD condition, 10 reported having had DSD-related surgery to the inner genitalia, outer genitalia, and/or gonads.

In **Study III**, HCPs were recruited to participate in the FGIs from September 2021 to November 2021. All HCPs who were familiar with DSD/intersex conditions and worked in collaboration with or within a multidisciplinary DSD/intersex team were invited to participate. Fourteen HCP participated in 3 FGIs during November 2021 and December 2021. The inclusion criterion for the FGIs was that the participants were familiar with the medical and psychosocial issues associated with living with a DSD/intersex condition.

The 14 HCPs had a variety of professional backgrounds, lengths of experience with DSD/intersex, and experiences with working in or in collaboration with one of the two regional multidisciplinary DSD/intersex teams. At the time of the FGIs, the COVID-19 pandemic was still ongoing, and some potential participants were prevented from participating due to illness or other COVID-19-related restrictions. Most of the HCPs worked with children up to 18 years of age and their families, although those with a gynecological or genetics background also worked with adults. The HCPs (seven females and seven males) were interviewed in focus groups and included in the data analyses in Study III.

Contact with Participants Prior to the Interviews

In **Studies I and II**, all the identified individuals living with a DSD condition received an information sheet about the study and a consent form (Appendix 4 and 5). Those who returned a signed consent form received a telephone call (in Paper I, from CH; in Paper II, from LM) to provide further information about the study and to arrange a time and place for the interview. The information sheet outlined the study's purpose and use of qualitative interviews, provided information about the interview topics, and included the main researcher's name and contact information. The sheet also contained key ethical information, such as the confidentiality measures and the participants' right to withdraw from the study at any time.

For **Study III**, the HCPs received an information sheet and consent form by e-mail (Appendix 6). The HCPs signed the consent form at the time of their interview.

Table 3

Demographic and Clinical Information Concerning the Individual Participants

	Adults Study I	**AYAs Study II**	**HCPs Study III**
Gender			
Female	10	8	8
Male	5	3	6
Age range			
16–20 years		5	
21–26 years		6	
30–49 years	12		7
50–70+ years	3		7
Time of diagnosis			
0–3 years	7	6	-
Pre-pubertal age (range: 4–12 years)	3	1	-
Adolescence (range: 13–18 years)	2	3	-
Adult (>18 years)	3	-	-
DSD-related surgery			
Yes	7	10	-
No	8	1	-

Table 4

Overview of the Conditions Represented in Individual Interviews

Diagnosis of Differences of Sex Development	Number
Congenital adrenal hypoplasia	8
Mayer-Rokitansky-Küster-Hauser syndrome	3
Hypospadias or structural associations of external genitalia	6
Turner syndrome	5
Klinefelter syndrome	2
Complete androgen insensitivity syndrome	1
Swyer syndrome	1

Data Collection

All the data were collected via in-depth individual interviews or FGIs. A thematic interview guide was developed prior to each study (Study I, see Appendix 1; Study 2, see Appendix 2; Study 3, see Appendix 3) to address the research purpose, as inspired by the scientific literature in the field (Kvale & Brinkmann 2009, p. 105) and with input from members of the two reference groups.

Qualitative In-Depth Interviews

Study I. The interviews with the adult participants aged over 30 years were conducted by a female researcher with a master's degree in psychology (CH) or by a female child psychiatrist (AW) who works in the field of DSD/intersex. Neither of them had met the participants prior to the interviews. The initial aims of this study were to explore the group's life situations and requirements for health and care services and interventions. These interviews were conducted on a face-to-face basis (n = 9) or by telephone (n=6), depending on each participant's preferences. The face-to-face interviews were conducted in meeting rooms located outside the clinic at the OUS in Norway, and the relevant participants' travel expenses were reimbursed. The interviews were audio recorded using a Zoom H2n Handy Recorder and then transcribed verbatim by CH and two research assistants.

Study II. The majority of the interviews with the AYAs were conducted by the Ph.D. candidate (LM), with one interview performed in tandem with a supervisor (LF) who has significant experience in the qualitative methodology and interviewing. The participants were made aware of my role as a Ph.D. student and told that I have clinical experience as a nurse working with children with DSD/intersex conditions and as a counsellor at a national

competence center for a variety of rare disorders. If I had previously met any of the participants in the course of my job, I informed them about it, even though they might not have remembered it themselves. I largely followed the lead of the participants in terms of the direction of the conversation. As sensitive subjects were discussed during the interviews, letting the participants take the lead gave them greater control over the discussion than if the interview guide had been followed in a strict manner. The interviews were conducted in the participants' homes (n = 2), in meeting rooms in the hospitals (n = 3), or via video (n = 5) or telephone (n = 1) due to COVID-19 restrictions. The participants' travel expenses were reimbursed when necessary. The interviews were audio recorded using a Zoom H2n Handy Recorder and then transcribed verbatim by me (n = 4) or a research assistant (n = 7).

The locations of the interviews were decided by the participants when possible. This meant that the physical prerequisites for each interview were quite different, which might have played a role in the knowledge production (Elwood & Martin, 2000). For instance, an interview with a young participant who was in town for a medical consultation took place in a meeting room at the hospital. Another interview was conducted by telephone. The participant had a hearing impairment, which led to some misunderstandings and laughter, whereas a face-to-face interview could have reduced the misunderstandings. Another interview was conducted via video, which seemed to give the participant some distance, making it easier to talk about very intimate details regarding the participant's sex life, genital appearance, and sexual health. As the locations of the interviews might have had different effects on the research process, it was important to be aware of such effects during the data analysis (Bjørvik et al., 2023).

FGIs

Study III. Two FGIs were conducted in meeting rooms in a hotel, and they involved two groups of four and five HCPs, respectively. The third FGI took place in a hospital and involved one group of five HCPs. Two of the focus groups were led by me and a psychologist with experience in conducting FGIs, while the third was led by two of the supervisors of the present Ph.D. project: AW, who is a child psychiatrist with experience of both DSD/intersex and group discussions, and SS, who is an experienced psychologist and researcher. Participants with similar characteristics but varied genders, professional backgrounds, geographical affiliations, and experiences with DSD/intersex were grouped together to discuss topics of common interest. This approach was applied to allow for diversity and ensure that there was room for different opinions regarding surgery, in addition to different ideas,

thoughts, and experiences regarding transitions. Each focus group was scheduled to last for between 60 and 90 minutes (actual duration range: 77–100 minutes), with plans to discuss two main topics: transitional needs when moving from child to adult health services and perspectives on DSD-related surgery. Consequently, the FGIs were more structured than the individual interviews, being divided into two parts. However, surgical perspectives were naturally touched upon when talking about transitions, and vice versa. After each FGI, field notes were written by the four researchers, and initial thoughts were discussed immediately after the interviews. The FGIs were audio recorded and transcribed verbatim by me. The transcribed audio recordings, field notes/reflexive summaries, and notes on the main impressions comprised the overall data collection tools. Moreover, each participant filled out a short form detailing their age, professional background, and experience with DSD/intersex prior to the interview.

Data Management

All the research data were stored securely on an IronKey encrypted USB storage device, which was protected by a password and locked inside a fireproof lockable container at OUS. The recorded data were transferred to a computer without an internet connection shortly after the interviews and prior to being transferred to the IronKey. Immediately after the transfer, recorded data were deleted from both the recorder and the computer. The deidentified interview transcripts were also stored in the IronKey. The data were only available to the PH.D. candidate and the main supervisor, while the de-identified transcripts were available to all three supervisors via access based on a secure login to the OUS server.

Data Analysis

The data analysis in this Ph.D. project was guided by the reflexive thematic analysis (RTA) approach, as described by Braun and Clarke (2006, 2019). RTA is a method able to identify patterns of meaning across datasets, enabling the elucidation of the research questions with the first-person perspective in mind (Braun & Clarke, 2006). Interpretation is embedded within the analytic process from beginning to end in the case of RTA. Interpretation is not about adding meaning to the data material; rather, it is about making sense, or finding the meaning, of the interviews (Braun & Clarke, 2021, p. 191). As RTA is theoretically flexible (Braun & Clarke, 2019), it represents a way of analyzing datasets that reflects the complexity of the phenomenon to be studied and understood.

Even though thematic analysis and the creation of knowledge represent a journey that starts with the formulation of research questions, the "formal" process of analyzing the dataset starts with familiarization with the material. In the present project, this included listening to and transcribing the interviews, taking notes, re-reading the transcripts, and trying to get an idea of what the participants were saying. The processes during study I, II, and III were a bit different, as I had conducted all the interviews in study II and III but not in Study I. This caused me to read and re-read the interviews several times to try to get a grip of the meaning, while simultaneously placing the interviews in context (done by phone, face-to-face, location, participant's age, who conducted the interview, etc.). After having familiarized myself with the interviews, I started the coding process. I aimed to put all the preliminary impressions of the interviews aside and inductively code the transcripts by hand. This means that I coded what the participants said, their own experiences, and their meanings (Braun & Clarke, 2021, p. 56). The codes were initially semantic, and close to the actual language used by the participants. When the analytic process developed and I added meaning and interpretation to the coding, a more latent level of coding was explored. After all the interviews were coded, I started organizing the codes into tables, where the initial candidate themes were created. Any irrelevant codes were grouped together as "miscellaneous" in order to ensure they were not left out. Subsequently, the candidate themes from the different interviews were compared, some new themes from across the interviews were created, and some themes where kept. The analysis process entailed a continuous process of going back and forth between the codes, themes, and subthemes. All the supervisors were involved in the analysis. Approximately 50% of the individual interviews were co-coded by one or more researchers. All three FGIs were coded by a minimum of two researchers (me and a supervisor). The codes were discussed, and the ideas were tested and re-tested until the final themes were clear. During this process, I checked the extracted quotes to see if the initial meanings had been changed or if other quotes described the themes or subthemes in a better way. This was necessary to ensure that the publications reflected the complex phenomena associated with living/working with DSD/intersex conditions.

Individual interviews. I did not discuss the same themes at length or in depth with all the participants. For example, most of the female participants discussed experiences or thoughts concerning fertility/infertility, although none of the men elaborated on this issue. As fertility was a topic we had initially identified as a key focus of this study, we had listed fertility in the interview guide. However, when the participants did not discuss this topic, I was careful about digging too deep into it, thinking it was not an issue for them and not

wanting to cause them additional worries, or maybe thinking they were unaware/uninterested in the topic due to their young age.

Some of the central themes in the interview guide (e.g., transition from child to adult among the AYAs) were not discussed in depth by the participants, while others that were not considered key areas of interest in the study were discussed in depth by several participants. Therefore, the themes presented in this study may be considered those of special concern for the participants rather than what we as researchers believed to be most important. The research methods facilitated this development. However, the themes discussed or raised by the participants may not have been the only ones significant to them. Factors such as time limitations, discomfort with bringing up sensitive dilemmas, and the interviewer's (the present researcher's) positioning in the field as a nurse who previously worked with DSD/intersex might have influenced this. Here, I had to respect the participants' silence, lack of elaboration, and apparent body language when trying to pursue some topics rather including what was not talked about in the data analysis.

Throughout the whole research process in general, and in the analysis in particular, it was important to place the stories related and the experiences described by the participants at the center of the thesis. By letting the participants guide the direction of the individual interviews, this study provides deeper insight into the areas regarded as important by the participants themselves.

Reflexivity

The present Ph.D. research project is an example of qualitative research with a reflexive approach whereby the researcher forms part of the knowledge construction alongside the participants and the rest of the research group (Braun & Clarke, 2019). In this section, a description of reflexivity in terms of personal matters will be provided, acknowledging what I brought to the project.

The purpose of writing this reflexivity section is to make explicit to both me as a researcher and the reader how I have influenced the steps of the research process (Olmos-Vega et al., 2023). The secondary goal of being reflexive is to enhance the transferability and transparency of the research process and findings (Braun & Clarke, 2019). As the primary researcher involved in this project, my professional background and work experience influenced the development of the project and will be elaborated on in this section.

I am a registered nurse who started my career working on a children's surgical ward in OUS with regional responsibility for the treatment of children with DSD/intersex conditions.

Taking care of the children, their siblings, and their families was a revelation. None of the families I was in contact with, as far I can remember, questioned the surgical solution to their "problem." Most of them saw it as a disease that could be treated with medicine and surgery. The same was true for most of my colleagues and for me as well. It took me a while to realize that life is not lived in the hospital and that individuals with rare disorders live meaningful lives outside the hospital, even though they are going through painful experiences when in the hospital. Later in my career, I started working at a national competence center for rare disorders. There, I met several children and adults with a variety of congenital malformations and with DSD/intersex conditions. They and their families came from all over Norway. These meetings took place on courses we arranged, during youth assemblies, or in meetings with their local community (e.g., schools, kindergarten, general practitioner, school-nurses, etc.). The main subjects the parents raised were issues regarding medication, how to talk about their child's condition within the family, how to handle gender-challenging behavior by a young child (e.g., children who identified as girls preferring to invite boys to birthday parties), and what to tell others. Due to these meetings, I was sensitized in terms of issues regarding disclosure, stigma, and dilemmas in both health care and decision making.

Despite having met individuals with DSD/intersex conditions and their families and caretakers in a variety of situations that most nurses do not encounter, I do not have an "insider" perspective. The participants regarded me as an HCP, not as a co-patient (and maybe not even as an ally). Some asked me about medical information (as I had disclosed my professional background), while others asked if I knew people who have the same condition as them. This facilitated an analytical approach from the outside, which is something it is important to be aware of. While an insider perspective might have offered advantages during the recruitment phase and in the interviews, some researchers have argued that an "outsider" perspective also offers benefits to the research process (Bridges & Bridges, 2017, p. 347).

The analysis was, whether I liked it or not, influenced by my privileged, reflexive position as a woman of White European ancestry, a nurse, a mother of teenagers, and a counsellor working in the field of rare disorders. This allowed me to use my lived experience of working with individuals born with DSD/intersex conditions and their families as a lens during the thematic interpretation of the interviews.

Ethical Considerations

Ethical considerations played a fundamental role in this project's research design and approach to knowledge. First, individuals with DSD/intersex conditions have historically been

subject to non-consensual treatment/medical investigation and a lack of disclosure concerning their condition. Moreover, they have been subject to treatment that neither they nor their caregivers fully understood. Thus, we incorporated a reference group to ensure that the aims of the study were relevant for affected individuals, that the language used in the interview guide was inclusive, and that the study design was transparent and inclusive.

Prior to conducting the study, ethical approval was obtained from the Regional Committee for Medical Research Ethics (Health Region Southeast, Norway, reference number: 79444) and from the Data Protection Officer at OUS (number: 7000898). Measures were taken to ensure that the research complied with the principles of the Declaration of Helsinki (World Medical Association., 2013). It was important to ensure that participation in the research was voluntarily and not a result of, for example, parental pressure. This was achieved by talking about the participants' motivation for participating and what they knew about their condition. A side effect of a well-informed participant is usually a more trusting relationship between the researcher and that participant, which can lead to better qualitative data (Tracy, 2010). Confidentiality was achieved by ensuring all personal data were de-identified and stored in a safe place. As Norway is a small country and as individuals diagnosed with DSD/intersex conditions in Norway are relatively few, measures were taken to safeguard the identities of the participants when reporting the data, including clustering ages and conditions, as well as removing or altering any identifiable information from the utilized quotes.

As mentioned above, the health-care field has a history of providing little information to patients in general, while parents might find it difficult to talk with their children about sex differences, meaning that individuals who receive an invitation to a study about DSD/intersex may face difficult questions or thoughts. The same is true for those who participated in the interviews. With this in mind, I was cautious when asking the participants if they could tell me about their conditions. If I felt that a participant knew very little about their condition, I tried to ask them carefully if there was something they wanted more information about or if they had any unanswered questions. If they wanted more information, an appointment with a suitable HCP was arranged for them.

Researchers are obligated to present the findings in a way that will not result in unjust or unintended consequences. HCPs have been subject to significant criticism from human rights activists due to previous and present practices associated with conducting surgery on children's genitalia before the age of consent (Carpenter, 2021). Prior to conducting FGIs with the HCPs about their experiences of DSD-related surgery, this issue was discussed in the

research group and with pediatric surgeons to ensure a balance between not presenting data in a sensational matter (which could further portray HCPs in a negative matter) and remaining true to the data. This entailed an interesting and very educational process in which I had to use reflexivity in a very conscious way. I asked myself questions such as "Why don't I use this quote?," "Why do I find this theme interesting?," "What is it about the way this participant is expressing themselves that makes me use more quotes from them?," and "Why am I hesitant to write about this theme?"

Interpretation and power. The process of interpretation poses ethical challenges because it involves the dilemma of telling a story that remains true to the participants' stories and, at the same time, not merely serving as a microphone for the participants' expressions. As interpretative RTA involves analyzing patterns of meaning by using the wider knowledge the researchers' bring to the table, the results will not necessarily be familiar to the individual participant, although there will be phenomena representing each interview/participant within the results (Braun & Clarke, 2021, p. 215). As this study involved questions of a sensitive nature, I had to be aware of the power I had when presenting the results. I had an ethical responsibility to be transparent during the analysis process and to ensure that the participants understood the purpose and nature of the study. Moreover, it was necessary neither to be patronizing when interviewing affected individuals nor to adopt an inferior attitude when interviewing or analyzing interviews with experienced HCPs. Braun & Clarke (2021, pp. 215–217) emphasizes how it is important to remain true to the obligations as a researcher and to be aware of the societal context, especially when representing persons from groups the researcher is not part of.

Literature Search

The literature search as part of this Ph.D. project was performed in collaboration with librarians from the medical library at OUS. The databases searched included Ovid Medline, PubMed, Cochrane Library, PsycInfo, and Google Scholar. In addition, manual searches and "snowball" searches were performed. The search criteria were studies involving individuals diagnosed with DSD/intersex conditions and studies published in either English or a Scandinavian language. The search strategies identified articles that featured the following words in their title, abstract, or main text: "differences in sex development," "disorders of sex development," "intersex," "stigma," "sexual health," "disclosure," "adolescents," "young adults," "adults," "health care professionals," "dilemmas," "surgery," and "autonomy."

Methodological Reflections

In this section, I will first reflect on some of the methodological choices made during this Ph.D. project — namely, recruitment, use of a reference group, the qualitative interviews, and the chosen path. Furthermore, I will discuss some of the central criteria when conducting qualitative research — that is, trustworthiness, triangulation, and transferability.

Recruitment

In Study I, the recruitment was performed by identifying suitable individuals via medical records, with the help of HCPs, via the voluntary registries of two competence centers for rare disorders, and through the member registries of four patient organizations (Klinefelter Association, Norwegian Association for Turner Syndrome, MRKH Norway, and National Association for CAH). For Studies II and III, the recruitment was performed through the registries of the two hospitals and via the voluntary registry of the Centre for Rare Disorders. These sampling techniques may have resulted in sampling bias, leading to an unwanted or hidden skewness in the data (Malterud, 2001).

Had we recruited participants via human right activists, the results might have been different. Such participants might have held different views on, for example, DSD/intersex-related surgery, disclosure, use of nomenclature, or experiences with the health-care system. The representativeness of the study is influenced by the recruitment phase; however, due to the rich descriptions provided of our own reflexivity, the society in which we conducted the research, and the participants' cultural background, the reader can draw their own conclusion regarding what happened in the research (Tracy, 2010).

The willingness to participate in research interviews may be influenced by several different factors. Individuals with DSD/intersex conditions may feel that they belong to a stigmatized group. Stigmatized individuals can be hard to reach, which is why we used the hospital registries for recruitment purposes (Barratt & Maddox, 2016). HCPs, a population often limited by their busy schedules, can also be hard to reach (Anthony & Danaher, 2016). Believing that a study is worthwhile may increase people's motivation to participate (Negrin et al., 2022). For instance, one participant in the present research expressed being motivated to take part by the opportunity to gain answers to some of his questions regarding his medical follow up as an adult, while another was motivated by being able to improve the care available for the younger generation. Others might have participated because they received invitations from the hospitals or were encouraged to read the information letter by a HCP they

trusted. Still, we managed to recruit fewer participants than planned, and the sensitivity of the topics involved might be one of the reasons for this (Giorgi, 2021).

We had few religious or cultural minorities represented among our participants. This represents a weakness of the research. However, even if we had managed to recruit more minority participants, we would still not be able to say that our study represents the experiences of a religious or cultural minority, as it is the individual experience we sought to understand (Braun & Clarke, 2021). In addition, this would have led to the even greater heterogeneity of the groups, making the analysis more complicated.

Reference Groups

The involvement of patient representatives is relevant to improving the quality and relevance of the research (Domecq et al., 2014). Furthermore, it is a criterion both for applying for research grants and for accessing funding from many agencies (Stiftelsen DAM, 2023). The insider perspective, which none of the members of the research group could provide, is what the reference groups contributed with. As mentioned above, the study involved two different reference groups. These groups had their own advantages and disadvantages. In Study I, there were few affected individuals involved. Out of ten participants, only one represented a patient group, while there were four individuals who represented a human rights activist organization. The input from the reference group was very useful, particularly during the initiation of the study, covering all the topics of interest. However, this represents another form of skewness, and the voices of the affected individuals might have been drowned out. As a consequence, the reference group in Studies II and III was composed solely of individuals with lived experiences of DSD. In addition, when writing Paper III based on the FGIs with the HCPs, we discussed the findings with three pediatric surgeons who had experience of working in the DSD/intersex field.

Reflection on the Qualitative Interviews

One challenge when using interviews as a method to obtain knowledge is that people tend to tell us what they think we want to hear, while their behaviors may not be consistent with their attitudes (Jerolmack & Khan, 2014). Jerolmack and Khan (2014) stated that when conducting interviews, researchers must be aware that it is not possible to analyze what people do, only what they say they think they do. Fortunately, when conducting a phenomenological study, the main focus is people's experiences and meanings.

When conducting interviews, it is necessary to be sensitive to the signals the participants are sending. For instance, is a given participant a bit tense? Is the participant trying to tell you what he thinks he wants you to hear? Does the participant seem well prepared and to have a story she wishes to convey? Is the participant open to the themes of the interview and does it feel more like a natural conversation than a research interview? All of these observations and experiences from the interviews form part of the analysis (Clarke & Braun, 2013).

The Chosen Path

We conducted the present research according to a phenomenological paradigm, using theory that illuminated the concepts of stigma, epistemic injustice, and healing to inform our theoretical framework and RTA as our methodology. This approach allowed us to explore a social process through the participants' and researchers' voices, and it also facilitated reflection on my role as both an insider and an outsider co-constructing data. Although we believe that this was the best methodological approach, it was certainly not the only option. Initially, a mixed-methods approach was considered to explore our research questions by including three questionnaires (WHO Quality of Life-BREF, Pediatric Quality of Life Inventory, and Strengths and Difficulties Questionnaire) that measure participants' QoL. When applying for ethical approval, the research group started to discuss the benefits of these questionnaires versus the strain they would place on the participants. As individuals living with DSD/intersex conditions in Norway represent a small population, we risked not being able to use the results of the questionnaires due to statistical issues. We also discussed whether both interviews and questionnaires would cause some potential participants to refuse to take part. After considerable discussion, the research group decided to focus on the theoretical framework of phenomenology. Being methodologically reflexive entailed understanding both the affordances and shortcomings of our choices and making them explicit in the final manuscript (Olmos-Vega et al., 2023).

Quality Criteria

The quality of the present research was tested using three criteria—namely, trustworthiness, triangulation, and transferability.

Trustworthiness. The concept of trustworthiness addresses whether a research method measures what it is intended to measure, in addition to whether the reader can trust that what they are reading is true (Clarke & Braun, 2013). To achieve trustworthiness, the

research needs to be situated culturally, in time, and in place. For instance, the results of our analysis revealed that very few of the participants questioned whether surgery on young children's genitals was ethically right or not. They accepted the treatment to be the best alternative due to their high level of confidence in the advice given by clinicians. If we had recruited participants via human right activists, the results might have been different and have displayed a more critical stance on the question. It is therefore important to provide rich descriptions of our own reflexivity, the society in which we conducted the research, and the participants' cultural backgrounds to ensure that the reader can draw their own conclusions about what happened in the research (Tracy, 2010). Still, it is important not to generalize, as there are always nuances that need to be investigated.

The research group comprised a variety of professionals with a diverse range of backgrounds. This was important to allow for the analysis and interpretation of themes from different perspectives and to ensure that the data were not misinterpreted (Tracy, 2010). This also helped us view the transcripts, interpretations, and results in different ways, to question our preunderstandings, and to gain a more in-depth understanding of the phenomena under study, thereby strengthening the trustworthiness of the research (Shenton, 2004).

A related factor that can strengthen the trustworthiness of a study is the positive effect of crystallization (Tracy, 2010), which is described as using different angles to view the research process. In our study, the research team was multidisciplinary (child psychiatry, neuropsychology, psychology, nursing, and cultural science), with the individual team members having more or less knowledge of the diagnostic group. This may have helped us view the transcripts, questions, and results in different ways. It may also have helped us gain a more in-depth understanding of the phenomena under study. In addition, feedback and collaboration with expert team members (in Studies I, II, and III) and clinical experts (as in Study III) helped us broaden our lenses when analyzing the material and ensured that the project was a worthy project that was relevant to the participants (Tracy, 2010).

Triangulation. Triangulation is used to develop a comprehensive understanding of complex phenomena (Malterud, 2001). It entails exploring a topic through different lenses, for example, by comparing the results obtained using two or more different methods and so broadening the understanding of the phenomena of interest (Triangulation, 2014). Qualitative and quantitative methods are complementary, and by using a mixed-methods approach, the knowledge of those living with DSD/intersex conditions could be expanded (Malterud, 2001). However, as we recruited a relatively small participant group, it was not realistic to achieve material that could confer sufficient sample power. Moreover, we did not feel that it was

ethically fair to subject the patients to the initially identified questionnaires, believing that the power would be too small to allow for publication of the results.

Another way to use triangulation involves obtaining data from two or more data sources (Malterud, 2001). In our project, we interviewed both AYAs, individuals over 30 years of age, and HCPs with longer and shorter lengths of experience with DSD/intersex. Consequently, the interviews and data analysis elicited rich information regarding personal experiences of living/working with DSD/intersex conditions from different data sources. That being said, I acknowledge that the project had some limitations, as the different interviews followed somewhat different interview guides and investigated different questions. Despite this, the phenomena of stigma, information sharing, and partial disclosure arose during both the individual interviews and the FGIs, strengthening the trustworthiness of the study.

Triangulation also demonstrates how other methods could have answered other questions in other ways. For instance, by initiating the project with a systematic literature review, I would have provided an overview of the qualitative and quantitative literature in the field. This would have been very valuable, both for the project and for me as a researcher. Furthermore, initiating an observational study in which I participated in the consultations where parents and patients received information about the condition and/or treatment path (including questions regarding DSD/intersex-related surgery) would have provided different information regarding, for example, the decision to perform surgery. Additionally, it would have further elucidated the dilemmas faced by HCPs, parents, and patients when making such decisions. However, there are both ethical and practical challenges associated with this approach, which stopped me from pursuing it.

Transferability. In terms of whether the findings of a research project can be used in another situation, transferability is achieved when the description of the study is sufficiently transparent and understandable that the reader can intuitively adopt the presented results in their own situation (Tracy, 2010). I have tried to be transparent about the different steps involved in the present research, describing the reflexivity and recruitment phase and providing detailed information about participants. However, the sensitivity of the topics under discussion made it difficult for me to be as transparent as I would have liked in order to provide the reader with the best starting point for situating the results and not compromising the principle of deidentification. The rarity of the conditions and the small population in Norway made this particularly difficult. The same was true for the description of the HCPs who willingly participated in the FGIs. We approached these ethical dilemmas by, for example, grouping the participants by both age and condition without revealing the rarest of

the conditions. Finally, the purpose of qualitative research is not to make the gathered data generalizable; rather, it is to contribute insights that can be applied in a given setting (Malterud, 2001).

Summary of the Results

Paper I:

"It was supposed to be a secret": A study of disclosure and stigma as experienced by adults with differences of sex development

Line Merete Mediå, Lena Fauske, Solrun Sigurdardottir, Kristin J. Billaud Feragen, Charlotte Heggeli, and Anne Wæhre

Published in *Health Psychology and Behavioral Medicine*

The importance of and need for full disclosure toward patients have been well documented. In the DSD/intersex field, the call for full disclosure was underscored in 2006 by a European multidisciplinary group of experts, which included patient representatives, due to the complexity of sexual development, the cultural impacts of DSD/intersex, and the sensitive nature of the information being shared. This group of experts also called for more research and knowledge regarding adults' experiences of living with DSD/intersex conditions, which could elucidate their everyday needs and improve the quality of the health-care services they receive over the course of their life. The aim of this paper was therefore to explore the everyday challenges faced by adults with DSD/intersex and to understand how issues such as disclosure, information sharing, and stigma affect their daily life.

To achieve these aims, 15 semi-structured interviews conducted with adults over the age of 30 years who had a DSD/intersex were analyzed via RTA. A phenomenological and hermeneutical approach was also applied to allow for the exploration of individuals' lived experiences and give meaning to those experiences. Adults with different DSD/intersex conditions were included to elucidate the similarities and differences associated with living with the different DSD/intersex conditions.

The main finding of this study concerned how adults with DSD/intersex struggle to achieve a balance between information sharing and concealment. Several described discovering for themselves that they were born with a DSD/intersex, indicating that they received too little information from both parents and HCPs when growing up, particularly in terms of sensitive and taboo subjects. This resulted in them not knowing how to talk to others about their DSD/intersex, in addition to not knowing how to ask for help when experiencing complications of their condition. Consequently, adults with DSD/intersex engage in invisible work to hide their difference, either by avoiding situations in which it could be revealed or

making preparations so that it is not revealed. It is important to recognize that this work is invisible to others, and it is often invisible to the participants themselves as well.

Several adults stated that children and adolescents require more and age-appropriate information when growing up, even though they may not demonstrate any interest. The results of this study also indicate that when individuals are diagnosed as adults, they experience difficulties grasping the implications of their condition, suggesting that the dissemination of information needs to be sensitive and customized even for adults. Thus, how related information is conveyed is important. Focusing on the medical aspect of the condition and how a person with a DSD/intersex condition differs from other persons may reinforce the feeling of stigma and being different. By contrast, focusing on common variations in human bodies and how DSD/intersex among these variations might help those with DSD/intersex to more easily accept their condition.

We conclude that individuals with DSD/intersex require lifelong multidisciplinary follow up and (renewed) information that is adjusted to their time of diagnosis, life situation, and psychological status.

Paper II:

Understanding sexual health concerns among adolescents and young adults with differences of sex development: A qualitative study

Line Merete Mediå, Solrun Sigurdardottir, Lena Fauske, and Anne Wæhre
Published in *International Journal of Qualitative Studies on Health and Well-Being*

This paper addresses the question of how AYAs living with DSD/intersex experience intimacy and sexual health. Previous studies and clinical understandings of these issues indicate that these experiences are complex and not easily measured, with contradictory results being visible in the literature. Moreover, the medical consequences of DSD/intersex (type and severity), psychological experiences of treatment (e.g., distress, anxiety), reactions from others (e.g., stigma), and lack of knowledge about sexuality have all been found to have a negative influence on sexual health. Among adolescents, questions about sexuality, identity, and intimacy become increasingly apparent, while having a chronic condition or being different in some way can affect how adolescents handle such issues. Consequently, the sexual, physical, and emotional health of adolescents, young adults, and adults may be affected.

This paper is based on empirical data derived from semi-structured qualitative interviews conducted with 11 AYAs aged 16–26 years who have been diagnosed with five different DSD/intersex conditions. We applied an interpretative phenomenological research design to achieve the aims of the study, and the interview findings were analyzed by means of RTA.

The overarching finding of this paper is the importance of being "normal," as described by the AYAs. It may seem that having a visible difference is more difficult, although having an invisible or concealable difference influences the lives of AYAs with DSD/intersex conditions. AYAs feel the need to engage in preparations to hide or to "normalize" their bodies before being intimate with others. Sex was described as problematic by both the male and female participants, who reported having genitals that were visibly different from the norm, resulting in them expending a lot of mental effort on feeling different. When AYAs lack knowledge about their bodily differences, it becomes difficult to communicate those differences to others. Moreover, not knowing why their body is different leads to feelings of insecurity that affect the participants' sexual relationships and result in a lack of everyday language with which to communicate about issues that concern them. This was also relevant in relation to questions regarding fertility. A lack of information or discrepancies in the information provided to AYAs are experienced as a significant strain. Interestingly, those who knew for sure that they could not get pregnant on their own reported having accepted their situation.

These findings are discussed in relation to theoretical understandings of how struggling to achieve normalization is a common coping strategy and form of stigma. Furthermore, this struggle entails adjustments and may affect AYAs' self-esteem. AYAs may expect stigma if they reveal their DSD condition and so may apply the coping strategy of hiding. By contrast, the findings show that after daring to reveal their differences, most AYAs received a positive reception from their partners.

The implications for practice of these findings include the need to recognize AYAs as a vulnerable group who require clear information, HCPs who are comfortable talking about sensitive issues, and the use of everyday language when communicating about their DSD. Moreover, support from peers is important for individuals with rare conditions. In conclusion, as being perceived as "normal" is important to AYAs, society should be more aware of the impact of how what is "normal" or common for AYAs in general, and for AYAs with DSD conditions in particular, is presented and discussed.

Paper III:

Dilemmas faced by health-care professionals regarding treatment and differences of sex development: A qualitative study

Line Merete Mediå, Lena Fauske, Solrun Sigurdardottir, Kristin J. Billaud Feragen, and Anne Wæhre

Submitted to *Health Psychology and Behavioral Medicine*

Paper III addresses questions regarding how HCPs experience the decision-making process associated with DSD-related surgeries and how the process is permeated by dilemmas. Prior studies have highlighted the numerous benefits of shared decision making, as well as the difficulties of making such decisions on the part of parents and affected individuals, although the perspectives of HCPs need to be further explored to better understand which dilemmas they face during this process and how to improve the health outcomes of individuals with DSD. To accomplish this, three qualitative FGIs with HCPs familiar with DSD were analyzed via RTA.

The findings of this paper are presented as the dilemmas faced by HCPs when initiating shared decision making with parents or individuals born with DSD. These dilemmas include questions about what considerations there should be, when surgery should be recommended, and when HCPs need to weigh different considerations against each other. One of the most challenging aspects was balancing caregivers' expectations regarding the outcomes of surgery against the child's right to make decisions about their body after reaching maturity. Subsequently, ethical dilemmas arise concerning what is in the best interests of the child—that is, to have normal looking genitalia or to have the possibility of having genitalia without the side effects of surgery. However, not all the HCPs considered this to be a dilemma. One participant stressed that parents who feel safe and comfortable with the situation can transfer their security to their child, whether having normalizing surgery on the genitalia or not. The identified dilemmas also include how to make decisions regarding DSD-related surgeries when HCPs need to deal with uncertainty and a lack of knowledge regarding the long-term outcomes of current treatment methods and when removing the functionally of gonads might pose a risk of cancer.

Taken together, these findings provide insights into the many difficult assessments that HCPs need to make as well as the choices that parents and/or affected individuals may have to make. The findings show that HCPs are genuinely concerned about providing the best possible care for children, adults, and their parents. Moreover, they reflect on the different

dilemmas regarding DSD-related surgery, although it is difficult for them to identify the best practice in many cases. Additionally, the fear of children, adolescents, and adults being stigmatized due to their differences may influence the advice given by HCPs.

The implications for practice here include the idea that HCPs should focus on affected individuals' and caregivers' reasoning and feelings when it comes to decisions regarding surgery, not on the severity of the relevant condition. The health-care community must acknowledge that dilemmas regarding DSD-related surgeries remain relevant today, some 17 years after the consensus statement was published. Consequently, research into the long-term consequences of undergoing or postponing DSD-related surgery for those affected by it is certainly warranted.

Discussion

Summary of the Answers to the Research Questions

The overall aim of this thesis was to explore the lived experiences of individuals with DSD/intersex conditions. The findings of Studies I and II, as derived from the in-depth interviews with adults and AYAs born with DSD/intersex conditions, and the findings of Study III, as derived from FGIs with HCPs, indicate that stigma influences several aspects of DSD/intersex. We found that individuals living with DSD/intersex strive to achieve a balance between concealing and revealing their condition, which influences the impact that DSD/intersex has on their life. Furthermore, issues related to communication, such as lacking everyday language with which to communicate issues to others and lacking knowledge, complicate the disclosure process. Even though few affected individuals describe issues concerning stigma in a direct matter, it is evident that the fear of not being "normal" has a major impact on individuals' lives, particularly when it comes to being intimate with others. Finally, decision making regarding DSD/intersex-related surgery is influenced by both the fear of stigma and the lack of evidence-based practice, as described by the HCPs.

In this chapter, I will discuss the findings of this Ph.D. project in relation to the theoretical underpinning of the thesis. When considering the findings of the three papers as a whole, the concept of stigma runs through the research and will serve as the theoretical background to the discussion. By focusing on the issue of disclosure, I will try to foster a better understanding of the lived experiences of individuals with DSD/intersex conditions through the concept of epistemic injustice. Finally, I will focus on how the concept of healing can elucidate lived experiences of DSD/intersex. The participants in Studies I and II represented a diverse group of individuals with a variety of lived experiences. However, there were both phenomena that were similar within the groups and phenomena that were different. Some of these phenomena will be explored in the following section.

In the interest of clarity, I will refer to the different studies by giving the relevant number or numbers in parentheses (e.g., (I) or (I and III)).

AYAs and Adults: Similarities and Differences

Studies I and II involved two different groups of participants. The first group comprised individuals with DSD/intersex conditions aged 30–70 years, whereas the second group comprised AYAs aged 16–26 years. Naturally, these two groups were diagnosed, treated, and followed up in different cultural times. However, all the participants (Studies I

and II) were born prior to the meeting in 2005 during which a consensus statement regarding the management of DSD/intersex was formulated and agreed upon by leading international professionals, patient groups, and activists in the field of DSD (Lee et al., 2006). I will now discuss some similarities regarding communication, knowledge, and understanding of different aspects of DSD/intersex, as well as some differences regarding perspectives on peer support, between the groups in light of the theoretical perspectives and previous studies in the field.

Similarities: Same Phenomena, Different Ages

In this project, a striking similarity was noted in terms of how the participants in Studies I and II described communication regarding DSD/intersex, lacking knowledge, and lacking everyday language. I had expected there to be a difference with regard to knowledge and understanding of their condition between the AYAs and adults, with the AYAs being better informed, particularly concerning how they described issues with communication after having been diagnosed and followed up in different times. Interestingly, the prevailing themes in both studies were difficulties with communication due to a lack of everyday language and a fear of stigmatization. However, very few participants described experiences with enacted stigma (i.e., stigma imposed by others; Goffman, 1963), although they still expected negative and stigmatized reactions from others. For example, Peter, one of the participants aged over 30 years (Study I), described how he does not participate in team sports due to having to use a communal shower and expose what he perceives as a different body. There may be several reasons for this. Choosing an open form of disclosure seems to have a positive effect (Bogart et al., 2022; Roth & Cohen, 1986). By contrast, those who experienced forced disclosure from friends or family members tended to experience this as stigmatizing (Bogart et al., 2022). Peter's situation might be understood as being forced to reveal his body in the shower, which causes him to avoid team sports.

During the interviews, we found that those aged over 30 years (Study I) who were diagnosed in childhood described being more open about their condition as they grew older when compared with individuals diagnosed in adolescence or as young adults. Receiving information about a condition that will affect sex development as an adolescent might be particularly difficult, as most AYAs wish to be like everybody else, meaning that they might potentially not be receptive to information provided by doctors. The concept of hermeneutic injustice may help us understand how this lack of receptivity and the associated knowledge gap make it harder for AYAs to make sense of their own lived experiences (Carpenter &

Jordens, 2022). In addition, those diagnosed in childhood (Study I) seemed to have accepted their differences, meaning they might have experienced less hermeneutic injustice. Furthermore, the adult participants (Study I) diagnosed in childhood experienced disclosure as more beneficial and found that explaining their condition to others could lead to normalization and reduced stigma. The results of Paper II shed light on this when demonstrating how a lack of understanding of one's own condition and the consequences of DSD make it more difficult to communicate with others (i.e., hermeneutic injustice). This can create a feeling of insecurity that results in a lack of everyday language with which to communicate, and it can also affect sexual relationships. This indicates that feeling like you have a stigmatized body and the fear of a stigmatizing gaze from others can, in certain situations, be time-dependent phenomena.

Another observed similarity related to the participants' desire for more, better, and customized information from HCPs. The same goes for the possibility of being a parent, the possible treatment options, how to have a bodily appearance like everybody else, and whether to stay diverse and learn to live with it. Previous studies have shown that AYAs believe HCPs to lack the tools necessary, or generally be reluctant, to talk about issues regarding sexual health (Callens et al., 2021). HCPs, including nurses, are in a unique position to communicate with individuals with DSD/intersex (Wisniewski et al., 2019). As DSD/intersex are rare conditions, HCPs may need specific communication skills and sensitivity to address the healthcare needs of affected individuals (Brennan et al., 2012; Wisniewski et al., 2019). For affected individuals to communicate about the difficult aspects of their diagnosis, they require appropriate everyday language. If HCPs use language that consists of mostly medical terminology, it suggests to patients that HCPs are the only ones they can talk to (Roen, 2019). This happens when testimonial injustice occurs and affected individuals assign too high a level of credibility to HCPs or too low a level to friends and family (Carel & Kidd, 2014). Consequently, the medicalized approach becomes prevalent, which may hamper affected individuals' communication with others.

Furthermore, evidence suggests that both affected individuals and the parents of children with DSD/intersex conditions describe how they "have" a condition, while they "are" not, for example, intersex (Feragen et al., 2019), which resonates with the results of Studies I and II. Prior studies have shown that "intersex" is a term few affected individuals or laypeople are familiar with, that few use the term "DSD," and that most use the name of the relevant medical condition (e.g., CAH or TS) (Feragen et al., 2019). Some even avoid repeating their diagnosis for fear of others googling it and discovering sensitive information about them or

their child (Lundberg et al., 2018). This uncertainty regarding certain central concepts associated with DSD/intersex can prevent affected individuals from broadening their knowledge when searching for information, for example, via the internet. If affected individuals and/or their families are unaware that there is a whole society of stakeholders and interest groups working toward the acceptance and normalization of DSD/intersex, it is not surprising that some affected individuals continue to believe that their body has to be changed in order to be defined as "normal", which may complicate disclosure.

Another perspective that can shed light on the practice of concealing DSD/intersex is the visibility of a given condition. Research has shown that individuals with visible differences (e.g., facial differences) fear that discussing those differences will lead to further stigma due to a lack of understanding of the cause and nature of their conditions (Bogart & Tickle-Degnen, 2015). A visible difference makes passing as a "normal" more difficulty (Myhre et al., 2021). However, while concealing certain differences can make it easier to fit in during specific situations, it can become even harder in other situations because revealing something unexpected about oneself can shatter people's preconceived notions. By contrast, Bogart et al. (2015) acknowledged disclosure to be an effective way of fostering understanding and advocating for those with visible differences. Some even actively educate others about their condition. The positive effect of disclosure was confirmed by van der Grift (2023) in individuals with DSD/intersex conditions. Their study compared the level of disclosure among individuals with different DSD/intersex conditions and demonstrated that women with virilization reported less openness, more shame, and more stigma, whereas women with TS reported the highest level of openness (van de Grift, 2023). The dilemma of choosing between concealing and revealing can lead to avoidance behavior, as described in a review of psychosocial health-care literature concerning individuals with DSD (Roen, 2019).

Differences: Experiences with Peer Support

What stood out as the difference in the findings was how the participants with hypospadias regarded peer support. In this context, peer support refers to a "mutually beneficial relationship in which persons having experienced or facing similar challenges share emotional, informational and social support" (Baratz et al., 2014, p.99). Peer support is acknowledged in the literature as one of the greatest healing tools and as leading to more understanding of an individual's own situation and strengths (Howe, 2021; MacKenzie et al., 2009; van de Grift et al., 2018). Thoits (2011) described similar others as individuals who represent a secondary group that enacts direct experiential knowledge and can provide

experienced-based support. In contrast to the primary group of significant others (e.g., family, close friends), who may be more emotionally involved due to the disruption the situation has caused to their own lives, similar others are less personally involved and can contribute peer support, act as role models, and allow affected individual to compare coping behaviors. Despite the positive documented effects of peer support (Thoits, 2011), evidence suggests that men with severe hypospadias tend to choose avoidance mechanisms as coping strategies (Bhatia et al., 2021; Örtqvist et al., 2017; Rynja et al., 2011). Most participants in Studies I and II talked in positive terms about meeting others with the same condition, and they wanted to serve as role models for newly diagnosed children or their parents. Conversely, unpublished material from the present project (I and II) reveals that some participants (males with hypospadias) do not want to meet people with the same diagnosis. They consider that disclosure has become easier with age, although when asked directly about this issue, they are reluctant to meet others. One participant's response to being asked how he felt about meeting others sheds light on this issue. He described the thought of meeting others with hypospadias as inappropriate and something he would never agree to. His description of this as "inappropriate" suggests the belief that his genitals and experiences are deviant in a negative way, meaning they are not something to be discussed with strangers, not even people with the same condition. Moreover, prior studies have shown that people with concealable stigmatized conditions are less likely to spend time with others with the same or similar conditions when compared to people with visible stigmatized conditions (Quinn & Chaudoir, 2015). By contrast, the individuals with DSD (and their parents) who participated in drafting the 2005 consensus statement valued peer support as a means of ending stigma and isolation, suggesting that a feeling of normalcy can be the result of children with DSD meeting peers (Lee et al., 2006).

Goffman (1963) claimed that stigmatized people prefer to be with people of their "own kind" rather than facing reactions in the "normal" world from unstigmatized people. Even though this finding is not in line with the perspectives concerning peer support related by our participants with hypospadias, it resonates with their openness during the interviews. The men with hypospadias were just as open as the other participants when it came to sharing intimate information, and they expressed that it felt good to talk to professionals. Another study based on in-depth interviews found that increased knowledge of an individual's diversity enables them to accept their own individuality (MacKenzie et al., 2009). The avoidance behavior demonstrated by our male participants with hypospadias (Studies I and II) may be an example of what Leder (2022) described as a healing strategy, where individuals combine ignoring and

objectifying their diverse body. It may be that affected individuals' distance themselves from having genitalia that look different from most others by incorporating a biomedical perspective—that is, "this penis is something I 'have', not something I 'am'." As the condition is not completely fixable, it should not be spoken about; rather, it should be ignored. At the same time, the men with hypospadias highlighted the need for more information during childhood and adolescence from HCPs and parents, in addition to more tools to enable them to practice disclosure (Paper I).

Previous studies have described how some individuals with DSD/intersex conditions choose to identify *as* intersex (Johnson et al., 2022; Lundberg et al., 2017; Monro et al., 2017) instead of being diagnosed *with* a DSD/intersex condition. This can be understood as being born with DSD/intersex being something "I am." Moreover, some individuals practice public disclosure, particularly those who are part of a peer support group or who have a human rights activist profile (e.g., InterACTadvocates.org). In Norway, we do not have a patient support group for individuals affected by hypospadias, nor are individuals born with hypospadias systematically offered to meet or speak to others with the same condition. This may add to the stigma associated with DSD/intersex in general and hypospadias in particular. In addition, most are not familiar with what DSD/intersex is when receiving the diagnosis of their condition (as a parent of a child with DSD/intersex or as a patient). This represents an example of epistemic injustice where parents or patients miss the groundbreaking knowledge of HCPs but risk only talking to HCPs about the condition, which again increases the medicalized view of DSD/intersex.

Based on the findings of Studies I and II, we suggest that the experience of living with DSD/intersex includes ambiguous elements and are colored by both similarities and differences. The fear of stigma complicates communication, and both AYAs and adults desire more information regarding their condition and its consequences. This can be understood as a consequence of the medical framing of DSD/intersex that prevails in health-care settings, meaning that some groups are reluctant to seek/do not have the option of peer support. Furthermore, the degree to which participants in studies I and II were comfortable with disclosure was influenced by their age and whether they were diagnosed in childhood or not. While some related positive experiences of having been open toward others, they still feared the same situation in other contexts. Studies have shown that disclosure has a positive effect on mental health, although it is still characterized by the fear of stigma in individuals with DSD/intersex conditions.

Normality

The concept of normality is prevalent in the field of medicine, as it represents the distinction between a diseased body and a healthy functioning body, which influences who requires diagnosis and/or treatment (Hofmann, 2005). Consequently, what is considered a disease is viewed as abnormal, whereas what is considered healthy is perceived as normal. The use of the word "normal" in the medical context can have a normative connotation, meaning that it sets the standard for how people should act or what the norms of medical treatment should be (Chadwick, 2017). The use of "normal" is also prominent in the DSD/intersex literature (De Clercq et al., 2022), where phenotypic sex is commonly described as either normal or abnormal, with hormones, genetics, and secondary sex characteristics being treated similarly. Furthermore, normalization, including striving for a sense of normalcy by portraying a normal life, has previously been identified as a common coping strategy among individuals with multiple conditions (Sanderson et al., 2011). In addition, cultural norms regarding the ideal appearance of a "normal" and attractive body can shape both a person's perception of their non-conforming physical appearance and the degree of disturbance of their sense of bodily identity (Eagly et al., 1991; Toombs, 1995). However, some studies have challenged the traditional conception of "normal" in relation to DSD/intersex, such as the work of Guntram (2013). Her study demonstrated how women who found out about their DSD/intersex condition as teens present themselves as "differently normal" (i.e., as slightly different from the norm) or "normally different" (i.e., a variety of normalcy).

Stigma is closely related to how DSD/intersex conditions are spoken about and how normality is framed (Hegarty & Smith, 2023). An example is when HCPs describe how surgery is a possibility for genitalia that looks different even when it is not medically necessarily (Roen & Hegarty, 2018). This narrative of how a body affected by DSD/intersex can look like most others with the help of surgery can contribute to a feeling of having the incorrect body and the belief that altering it via surgery is a way to "pass" as normal. This can be described as soul surgery, which has been defined as "surgery of the body to maintain primarily mental outcomes" (Hofmann, 2022, p. 1). It can also be an example of "social surgery," which is surgery performed to obtain primary social outcomes such as confirming social constructs of what male and female bodies should look like (Hofmann, 2022, p. 2). Hofmann (2022) noted that when the outcome of surgery is no longer dependent on the results achieved in the operating room, instead being dependent on the effect on the individual's mental health in the future or on social outcomes, new perspectives must to be considered.

"Soul surgery" reveals a problem highlighted by researchers when surgery performed on infants is intended to prevent psychological distress, despite evidence indicating that this connection is difficult to achieve (Hart & Shakespeare-Finch, 2022; Liao et al., 2019). This resonates with the findings of Study III, where the HCPs acknowledged that there is a lack of evidence concerning both the costs and the effects of, for example, feminizing surgery, although the expectations of some parents regarding normalizing their child's genitalia are strong and result in dilemmas for decision makers. Another problem associated with "social surgery" is that the alteration of the body via surgery continues to support the norm of how a body should be or look. This resonates with the dilemmas discussed in relation to DSD-related surgery in the present study. As few adults have not undergone "normalizing" surgery on their external genitalia, both HCPs and affected individuals and their parents continue to perceive such surgery as the norm. This may continue to be the case until the children of today who have not been operated on grow up and address this subject. A complicating factor is that there is no systematic, national follow up of adults with DSD in Norway, meaning that knowledge in DSD-related surgery is lacking. Based on this, it is important to introduce a national high-quality registry so that the potential to surveille outcomes is greater. This registry, if linked to European and international registries such as the "I-CAH" and "I-DSD" registries, has the potential to improve care of people with rare conditions and diseases, including heterogeneous conditions such as DSD/intersex (Kourime et al., 2017).

In addition to the obvious effect of hermeneutic injustice (i.e., that HCPs have more knowledge of rare and chronic conditions than most people do), we must consider how testimonial injustice effects communication between HCPs and affected individuals and their families (Fricker, 2017). For instance, while HCPs generally intend to provide thorough information about the pros and cons of female genital surgery, an HCP in Paper III described how this might be viewed as an argument *for* surgery. The HCP further explained how some parents argue strongly for their infants to have surgery and how HCPs sometimes accept such arguments reluctantly despite the consensus statement recommending a more cautious approach to early and medically unnecessary genital surgery intended to "normalize" the appearance of genitalia. Other studies that considered parents' wishes indicated that they not always feel that the choice regarding surgery was really theirs, given that they merely followed recommendations due to assigning high credibility to HCPs (Carel & Kidd, 2014). This further indicates that the expected stigma and the anticipation of being normal are strong. In Study III, some HCPs described how they found it particularly difficult to manage parents' clear wishes for early genital surgery to be performed on their child.

The present Ph.D. research project found that the fear and/or expectation of stigma influence the lives of both AYAs and adults living with DSD/intersex conditions. The results of Study III, as derived from FGIs with HCPs, also indicate that patients' expectations of being stigmatized influence their choices regarding DSD-related surgery. It is commonly believed that HCPs perform normalizing surgery to ease the psychological burden of both parents and children with DSD/intersex (Wisniewski, 2017). Yet, surgery performed with the intention of making a body "normal" and establishing wholeness for an individual with DSD/intersex can have the opposite effect and disturb the individual's bodily image. This can be experienced in a physical way when the person who has undergone genital surgery can feel the scar tissue with his fingers, which creates a disruption to his body image rather than a sense of wholeness because the body is more "normal." It can also be experienced in a sensational way when the gaze of a sexual partner on the altered appearance of an individual's genitalia can give the affected person the feeling of a "lump" in the stomach due to worry about being rejected, which corresponds with the experience of expected or experienced stigma (Goffman, 1963). This is illustrated in the case of Agnes (Paper II), who reported experiencing pain during sexual intercourse but explained that having nice-looking genitals is more important because that is what her partner sees. Agnes has undergone surgical correctional treatment, and she wants even more surgeries to be able to pass as "normal" and to not have stigmatized traits/be a stigmatized person.

This thesis does not aim to resolve the issue of what is the right thing to do when it comes to DSD/intersex-related surgery. Should we offer "normalizing" surgery or should we follow the approach of other countries and ban these elective surgeries? In phenomenological research, one should primarily describe and try to understand the lived experiences of individuals, and our focus was on understanding the experiences of affected individuals and HCPs (Kvale & Brinkmann, 2009). Some commentators have suggested that DSD/intersex-related surgery is performed to help parents accept their child's difference, in addition to improving the esthetics, functioning, and psychosexual development. Moreover, it has been suggested that such surgery is not beneficial for the affected person because it may cause trauma and decrease the sensitivity of the genitals (e.g., Diamond & Garland, 2014; Hart & Shakespeare-Finch, 2022; Roen, 2019). Such arguments are only partly supported by the findings of other research studies (Bennecke et al., 2021) and the data in this thesis. The participants in Study II accepted surgical interventions as a means of becoming more "normal," and some expressed the desire for more surgery. Even though these opinions were not expressed in the same way by the participants in Study I, the importance of having a

normally functioning and looking body, and of not problematizing the surgery that several of them have undergone, indicate that their attitudes toward DSD/intersex-related surgery might be similar to those of the participants in Study II.

Leder's (2022) healing strategies accord with the shared decision making of our participants, including healing by objectifying the body as something that can be fixed. Different groups of people and different individuals both emphasize and de-emphasize a diversity of things (e.g., importance of fertility, appearance) at different times (acceptance in school, impact on sexual health as adults). This demonstrates how they strive to overcome the ambivalence of accepting and changing the body, and it also reveals how an individualized approach when dealing with a DSD/intersex condition is beneficial. When the AYAs with DSD/intersex conditions talked about whether the sexual act was successful, it appeared to depend on their partner's reactions. This could be a healing strategy that helps AYAs to cope with their fear of intimacy and sexual activity through ignoring their different body and focusing on their partner's response (Leder, 2022).

The participants in our study sought to achieve normality in different ways, for example, by surgically improving a different body image, by not being "discovered," and by being accepted as they are. While none of the participants described receiving a "disapproving gaze" or hurtful comments from HCPs, the consequences of having a functionally and/or visibly different body might lead to them feeling different.

Shared Decision Making

We found that shared decision making (SDM) was a central concept in Study III. SDM—that is, a process of acknowledging that a decision is required, knowing and understanding the best available evidence, and incorporating the patient's values and preferences into the required decision—is a well-known tool for improving treatment agreement between patients and clinicians through building consensus and sharing information (Légaré & Witteman, 2013). Decisional aids have been developed for clinicians, patients, and extended family members, and they can take the form of e-learning, paper-based information, personal coaching, or mobile applications. However, these decisional aids are less frequently used in relation to surgical decision making (Niburski et al., 2020). The process of SDM is well suited to DSD/intersex-related surgeries because the time available to make a decision is not scant, the process itself is debated, and the surgery is often irreversible. Therefore, decisions should be made based on the best possible grounds. In addition, patients and clinicians should consider non-surgical alternatives as part of the decision-making process

(Niburski et al., 2020). A meta-analytical review of the literature describing SDM in relation to surgery reported a decrease in surgical rates, an increase in knowledge, and an increase in decisional satisfaction (8 out of 11 studies) when SDM tools were used (Niburski et al., 2020). This was confirmed by a review demonstrating that SDM "reduced the number of people choosing major elective invasive surgery in favor of more conservative options" (Stacey et al., 2017, p.2).

The participants in Studies I and II did not question medical and surgical treatment being performed during childhood. By questioning the speakers beliefs, researchers risk perpetuating epistemic injustice by not assigning credibility to the speaker and interpreting this to fit our "prejudice" that individuals with DSD/intersex are not able to make up their own mind based on a well-informed knowledge base (Fricker, 2017). Merrick (2019, p.4435) noted that there is a "marginalization of the female voice and perspective, a long standing problem in western biomedicine and much discussed topic in feminist philosophy of science." Thus, we should perhaps give more credit to the voice of females who claim that they are satisfied with DSD/intersex-related surgery performed in childhood. Binet (2016) demonstrated that 90% of participants (n = 16) with CAH who had undergone surgery during childhood tend to maintain in adult life that surgery should be performed in infancy.

However, ethical considerations must be taken into account when the question of whether parents of young children with DSD/intersex conditions should be allowed to make elective and irreversible surgical decisions, particularly when evidence of the risks or positive effects is scarce (Sandberg & Vilain, 2022). It has been emphasized that there are challenges associated with systematically gathering data among small populations with a diverse phenotype due to issues such as sample bias (from patient associations). Moreover, there may be bias due to researchers serving as clinicians, while there may also be a lack of control groups and a lack of standardized measurement instruments (Lux, 2009). Conversely, we must consider how a ban on DSD/intersex-related surgery may impact a child's well-being (Sandberg & Vilain, 2022). In light of these issues, multidisciplinary teams could benefit from addressing the identified ethical considerations more actively and including medical ethics in the treatment of affected individuals (Lee et al., 2006).

Hermeneutical injustice, which occurs when patients do not have a concept with which to articulate their experience of illness, also accords with the issue with SDM raised by our participants. In Study I, the participants reported lacking knowledge and everyday language, which decreased their level of credibility. For example, one participant described an episode from her childhood where the decision regarding the introduction of hormonal shots to

postpone pubertal development was taken without including her perspective. HCPs assumed that they knew what was in her best interest (testimonial injustice). Another example we can dwell on is the lack of systematic information about how to contact peers and the lack of facilitation of such contact. If this unwillingness to create a link with peers is due to HCPs believing such contact and knowledge to be irrelevant, unfavorable, or damaging, the HCPs are practicing testimonial injustice, which might cause patients and their families to miss the opportunity to meet peers for support (Carpenter & Jordens, 2022). Furthermore, prior reports have revealed that trust issues might exist between HCPs and those who provide peer support that goes both ways, limiting the provision of systematical peer support to, for example, newly diagnosed individuals (Lossie & Green, 2015).

Another bioethical perspective concerns whether HCPs assess anecdotal qualitative information or qualitative research as equally reliable when compared with quantitative research. In Study III, the issue of whether activists' voices provide merely anecdotal statements that cannot be considered evidence in support of banning DSD/intersex-related surgery was debated. In the same interviews, testimonies from satisfied adolescents were taken into account as evidence of hypospadias surgery being positive. This prompts us to question which voices are listened to and where are the testimonies from satisfied adults? These ethical issues need to be further explored due to intersecting with principles related to patient autonomy, equity, informed consent, and the pursuit of high-quality care. While qualitative data may not offer the same statistical precision as quantitative data, it can provide valuable contextual information and insights that are crucial for ethical decision making (Braun & Clarke, 2019).

Strengths and Weaknesses

The primary strength of this study lies in the unique and rich in-depth data gathered on personal experiences of living with DSD/intersex, as related by AYAs and adults born with a DSD/intersex condition, a previously understudied group of individuals. To the best of our knowledge, the Bufdir project together with the present study are the only studies to involve interviews with AYAs and adults aged 30–70 years as well as FGIs with HCPs who work with DSD/intersex in Norway. We recruited participants of varying ages, phenotypes, and demographic backgrounds in an attempt to capture to the greatest extent possible the variation in experiences and increase the dependability of the research (in Study I participants were selected strategically). The participants also reflected both current and prior lived experience of DSD/intersex. However, this research presents the experiences of participants who, in

different ways, received an invitation to participate via channels through which persons with DSD/intersex conditions typically receive formal or informal health-care services and/or support. The findings must therefore be considered within this context. For example, the participants may include a high number of individuals who are only used to a medical framing of DSD/intersex, meaning that they may not have been informed of, or be interested in, a more human rights activist framing of DSD/intersex. Consequently, other AYAs and adults with DSD/intersex conditions may have different experiences when compared with the lived experiences conveyed in this research.

Another strength of this research, which may also prove to be a limitation, is the perspective that I brought to the project. As described in the reflexivity section, I have an outsider perspective. Still, my perspective is founded on a diversity of experiences with issues regarding DSD/intersex (see page 39-41). When interviewing the AYAs, the age discrepancies between us could have enhanced the power disparity. Conversely, it could have been a strength if they considered talking to a researcher nearer to their own age to be confining.

Looking back at what we have learned in this study, the fact that the expectation of there being a difference between the AYAs and adults, including a diversity in terms of age (AYA, adults, and elder adults), was determined to be unfounded represents another strength of this research.

A limitation of this study relates to the fact that the interviews did not cover sexual activity in much detail (e.g., frequency, type of activity, partner's gender). As the participants guided the direction of the interviews, some talked in more detail regarding sexual activity, whereas others were hesitant to discuss this issue. As some AYAs may be unwilling to talk in detail about sexual activity unless requested to do so, future studies could facilitate such discussion when ethically appropriate through preparing the participants during initial conversations or via the invitation letter and so avoid a two-way taboo.

Prior to Studies II and III, we did not perform a pilot interview. This may represent a weakness of this research, as it would have left us better prepared for the rest of the interviews. The reason for this omission was the fact that the rarity of the conditions may have posed a challenge in terms of recruiting patients (as we later found it to be). As a consequence of lacking a pilot interview, we discussed the interview guide after the first interviews and revised both the formulation of the questions and the emphasis on the main topics raised by the participants.

Conclusion

To better understand and help those born with DSD/intersex conditions, HCPs must comprehend the individual patient's experiences as well as how they assign meaning to those experiences. This Ph.D. research project represents a contribution to the ongoing exploration of the complex dynamics through which social, psychological, and biological processes combine to influence health. Consequently, this thesis extends the knowledge in the field of DSD/intersex and has the potential to improve healthcare for individuals living with DSD/intersex conditions.

This thesis provides a deeper insight into experiences of people living with DSD/intersex conditions as well as those of HCPs who work in the field. More specifically, this thesis makes three important contributions to the field and to the phenomenology of lived experiences of DSD/intersex. First, through interviews with adults aged up to 70 years, we learned how experiences of growing up with a DSD/intersex condition are influenced by stigma and how the issue of disclosure persists as an important aspect of their life during adulthood. Second, through interviews with AYAs, we learned of the importance of being "normal" and the influence that a concealable stigmatized condition can have on both intimacy and sexual health. Finally, through interviews with HCPs, we learned how the decision-making process regarding DSD/intersex-related surgeries is permeated by dilemmas. The lack of evidence-based knowledge, the expectations of affected persons and their parents, and the notion of a child's best interest are all linked to DSD/intersex being stigmatized conditions. This thesis highlights the challenges associated with a medicalized hegemony in the field of DSD/intersex, with both social and cultural norms affecting expectations of stigma.

Moreover, this thesis makes several important contributions to the body of Norwegian public information. For example, it is necessary to make people more aware of the issues relevant to people with DSD/intersex conditions, and to ensure that the related conversation takes place in language that is accessible to the public, if the debate concerning the pros and cons DSD/intersex-related surgery is ever to reach a consensus. The findings presented in this thesis suggest that it is the desire to feel "normal" that drives individuals more than anything else—the wish for a body that will "pass as normal," one that we and others can accept, enjoy, embrace, and even love—meaning that such feelings might be far more important than the condition itself. By speaking to the public about the diversity of genital appearances, the term "normal" might achieve a broader understanding.

Finally, it appears that living with a DSD/intersex condition impacts individuals both mentally and physically. The possibility of receiving psychological consultation should therefore be offered as a rule, not merely available as an option or something that must be requested. For some, psychosocial interventions would preferably take place in their local community, as they live some distance away from their multidisciplinary DSD/intersex team and often attend once a year.

Implications

The findings of this Ph.D. research project have implications for affected individuals, their families, HCPs, and society in general. As those living with DSD/intersex conditions face the risk of experiencing stigma, society's overall level of knowledge of bodies with congenital conditions that diverge from the norm of what is expected of a male or female body should be enhanced. In Norway, the debate concerning whether there are more than two genders (a biological perspective), and also concerning transgender acceptance and rights, has received significant attention in the public discourse, although narratives and discussions regarding DSD/intersex are still almost non-existent. The Center for Rare Disorders, a national competence center located within Oslo University Hospital that is tasked with collecting, consolidating, and spreading expertise on rare conditions such as DSD/intersex, should contribute to this and help make DSD/intersex better known.

Moreover, we aimed for transparency in terms of the description of this work, and the findings of this Ph.D. research project can consequently be transferred to other concealable conditions, such as living with a stoma or with burns.

The results presented in Papers I and II indicate the need for better planning regarding the transition from child- to adult health-care services, where children and AYAs should receive information about their condition and training in talking about it when desired and required. This should be facilitated by HCPs working alongside parents, and it should serve to make the transition to adulthood easier.

The findings discussed in this thesis also suggest that stigma has a strong influence on the lives of those living with DSD/intersex conditions. Thus, HCPs, affected individuals, and their families all need to be aware on the effect that stigma can have and to acknowledge that effect in health-care settings.

Finally, the interviews with HCPs conducted during the course of this research highlight the need for more resources to be dedicated to researching the consequences of the DSD/intersex treatments currently offered to patients.

Suggestions for Future Research

The findings of this thesis raise several important questions that could be addressed in future studies. As part of the original project plan, we aimed to explore the needs of AYAs when transitioning from a children's department to an adult department. One promising avenue for future research might be to include the implications of sexual health, stigma, and lack of everyday language when testing an intervention to develop evidence-based guidelines for this transitional phase. Furthermore, Leder's (2022) concept of healing, as discussed in this thesis, could serve as a useful framework for understanding the needs of AYAs during this transitional phase.

Furthermore, more research on adults living with DSD/intersex conditions is required to elucidate the consequences of current practice in this field. The results of both the present Ph.D. research project and previous research highlight the lack of evidence-based knowledge that can serve as a backdrop for medical and psychological practice. We need to systematically follow up both those who have been treated as young children and those who have not undergone surgery on their genitalia to undercover both groups' follow-up needs and ensure they have effective access to appropriate health care throughout their lives. The focus here must be multidimensional, including both medical and psychosocial needs. In addition, the parental perspective is invaluable when it comes to understanding shared decision making, openness, and the impact of stigma, although it still requires further exploration.

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Paper I-III

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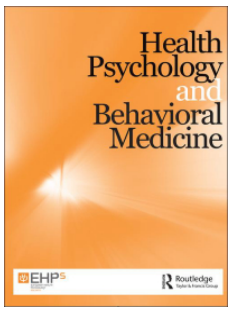

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'It was Supposed to be a Secret': a study of disclosure and stigma as experienced by adults with differences of sex development

Line Merete Mediå[a,b], Lena Fauske[b,c], Solrun Sigurdardottir[a], Kristin J. Billaud Feragen[a], Charlotte Heggeli[a,d] and Anne Wæhre[e]

[a]Women and Children's Division, Centre for Rare Disorders, Oslo University Hospital, Oslo, Norway; [b]Department of Interdisciplinary Health Sciences, Institute of Health and Society, University of Oslo, Oslo, Norway; [c]Department of Oncology, Norwegian Radium Hospital, Oslo University Hospital, Oslo, Norway; [d]Educational Psychology Service, Tonsberg Municipality, Tonsberg, Norway; [e]Department of Child and Adolescent Psychiatry, Oslo University Hospital and Institute of Clinical Medicine, Oslo, Norway

ABSTRACT

Background: Differences of sex development (DSD) are a group of congenital conditions that involve variations in sex chromosomes, genes, external and/or internal genitalia, hormones, and secondary sex characteristics. The present study sought to highlight the everyday challenges faced by adults with DSD as well as to understand how issues such as disclosure, information sharing, and stigma affect their daily life.

Method: We applied an interpretative phenomenological study design to explore the first-person perspectives. Semi-structured qualitative interviews of 15 adults aged 30–70 years living in Norway with five different DSD conditions (Turner syndrome, Klinefelter syndrome, congenital adrenal hyperplasia, Mayer-Rokitansky-Küster-Hauser syndrome and hypospadias) were analyzed using reflexive thematic analysis.

Results: Living with DSD, indicated doing a balancing act between hiding and/or exposing what participants perceived differed from others bodies. Communication regarding sensitive topics proved to be important. The participants were doing invisible work to manage the balance between concealing and revealing their feeling of differentness, a work effort that was not necessarily perceivable to others but still affected everyday life of the participants. Furthermore, the participants' experiences of disclosure changed over time, as those who were diagnosed during childhood found that disclosure became easier with advancing age. However, being diagnosed as an adult seemed to increase the feeling of difference and complicate disclosure.

Conclusion: Individuals with DSD should receive adequate information and have someone to practice disclosure towards, which could possibly strengthen the psychosocial aspects of living with their condition. The results emphasize the need to

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CONTACT Line Merete Mediå lmedia@ous-hf.no Women and Children's Division, Centre for Rare Disorders, Oslo University Hospital, 0373 Oslo, Norway; Department of Interdisciplinary Health Sciences, Institute of Health and Society, University of Oslo, 0373 Oslo, Norway

help individuals with DSD achieve a balance between disclosure and self-protection, overcome stigma, and determine when and how information about their DSD should be provided to others.

Introduction

Differences of sex development (DSD), which are also referred to as disorders of sex development or intersex, are a group of conditions that involve variations in individuals' sex characteristics, resulting in their genitals, hormones, or chromosomes differing from traditional conceptions of male and female bodies (Lee, Houk, Ahmed, & Hughes, 2006). The DSD population is large and heterogeneous with regard to diagnoses, severity of medical complications, psychological impacts, treatments, and follow-up (Kim & Kim, 2012; Lee et al., 2006). Some patients have visible variations in phenotype (e.g. in their genital appearance), while others exhibit differences in genotype (e.g. in their sex chromosomes). The estimated incidence of DSD varies from 1:200–1:300 (García-Acero, Moreno, Suárez, & Rojas, 2019), to 1:4500–1:5500 newborns (Sax, 2002). DSD are complex conditions that affect not only physiological processes within the body. Any chronic health condition can potentially affect the sense of identity and psychological well-being. In addition, treatments and the way others respond to bodily differences can have a negative impact on mental health in individuals with DSD. Knowledge and understanding of these impacts are important for those of us born with DSD.

In 2006, a consensus statement concerning the management of DSD was published, recommending that the evaluation and long-term care of people affected by DSD should be conducted at medical centers with multidisciplinary teams familiar with their medical needs (Lee et al., 2006). In Norway, multidisciplinary teams follow-up children with DSD until the age of 18. After the age of 18 years, there are no organized multidisciplinary follow-ups. Multidisciplinary DSD teams can provide psychological support to affected individuals and their families as a standard component of care, with children receiving age-appropriate medical information, and gender issues being discussed (Hiort et al., 2014).

The debate concerning the level of disclosure towards affected individuals, information sharing and multidiscipline follow-up of adults is not new within the field of DSD (Sutton et al., 2006; Tremblay, Van Vliet, Gonthier, & Janvier, 2016). Already in the 1950s, the debate about autonomy and shared decision-making, which is rooted in well-informed patients regarding their condition and bodily difference, was central as a consequence of the Nurnberg codex and the Geneva declaration (Reis, 2019), a debate that was ongoing for decades. In the 1980s, John Money (1987) wrote on the importance of educating patients about their condition in ways that might reduce the likelihood of it being received as stigmatizing. Stigma stems from undesirable attributes that people typically seek to avoid (Goffman, 1963, pp. 3–4).

In 2006, the consensus statement underscored the importance of full disclosure toward individuals with DSD (Howe, 2021; Lee et al., 2016). However, there still remained challenges in terms of sharing information about DSD, given the complexity of sexual development, the cultural impacts of DSD, and the sensitive nature of the information being shared (Lampalzer, Briken, & Schweizer, 2021; Malmqvist & Zeiler, 2010; McCauley, 2017; Weidler & Peterson, 2019). Moreover, advice concerning whether or

not to share such information with a wider circle of people was recognized as a complicated issue (Hughes, Nihoul-Fekete, Thomas, & Cohen-Kettenis, 2007). In addition to challenges concerning disclosure, individuals with DSD may experience stigma in social contexts that may increase their communication difficulties (Earnshaw & Quinn, 2012). Recent studies have examined the effects of stigma on people with DSD and identified both feelings of shame (i.e. experienced stigma) (Engberg, Moller, Hagenfeldt, Nordenskjold, & Frisen, 2016) and withdrawal behavior (i.e. anticipated stigma) (Meyer-Bahlburg, Khuri, Reyes-Portillo, & New, 2017; Meyer-Bahlburg, Khuri, Reyes-Portillo, Ehrhardt, & New, 2018).

Qualitative research that gives voice to the individual experience and provides a more in-depth understanding of the everyday life of people with DSD is scarce (Lundberg, Donasen, Hegarty, & Roen, 2019; Roen, 2018). In particular, the 2006 consensus statement highlighted the need for research on adults' experiences of living with DSD that could elucidate their everyday needs and improve the quality of healthcare over the course of life (Cools et al., 2018).

Given that DSD can negatively affect individuals' psychosocial well-being and quality of life (e.g. due to a lack of information, difficulties with disclosure and stigma), there exists a pressing need to identify areas of life in which affected individuals might require additional support. In order to make greater use of valuable resources in the health care system, clinicians need to investigate adults` personal experiences of living with DSD. The overall aim of the present study was to describe the everyday challenges faced by adults with DSD and to explore how issues such as disclosure, information sharing, and stigma affect their daily life.

Materials and methods

To achieve the aims of this study, we applied a qualitative and explorative design, and utilized a phenomenological and hermeneutical approach. Phenomenology is a systematic examination of different ways of experiencing reality. It seeks to explore and understand the lived experience of a phenomenon, and the way it is experienced and described by the individuals themselves (Kvale & Brinkmann, 2009). A hermeneutical approach implies a method used to understand and interpret the phenomena as expressed by participants (Kvale & Brinkmann, 2009). The approach is based on the participant's and the researcher's preunderstandings, on the context of the interviews, and develops throughout the entire research process (Gubrium, Holstein, Marvasti, & McKinney, 2012). We sought to understand how the participants described their experience of living with DSD, and how they perceived and spoke about their diagnosis to others by using semi-structured interviews as a tool.

We chose to include different diagnoses of DSD to shed lights on what was similar, or what distinguished the different diagnoses in adults aged 30–70 years.

Participants

The total number of participants comprised a convenience sample of 15 adults. All participants expressed identifying with the sex they were assigned at birth (five males and ten females). They differed with regard to their diagnosis, age (range: 30–70 years, M_{age}: 44

years), time of diagnosis made (children, adults), and number of surgeries. An approximate age range is reported for the purpose of de-identification. In the following, we present demographic descriptions for each diagnostic group represented in the material based on information from the participants in the semi-structured interview: Five of the female participants had Turner syndrome (TS). Of these, four were diagnosed within pre-pubertal age (range: 4–12 years) and one was diagnosed as adult. The group reported no surgical interventions related to DSD. Three female participants had congenital adrenal hyperplasia (CAH), and all were diagnosed within the first five years of life. In this group, all had undergone surgery on the genitalia within the first two years of life. Of these, two had undergone correctional surgery in pre-pubertal phase and/or as young adults. Two female participants had Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) and were diagnosed in adolescence (range 14–18 years). Of these, one had undergone two operations, including a vaginoplasty, and the other had none. Three male participants had proximal hypospadias (referred to as hypospadias in text). They were all diagnosed within the first two years of life and had undergone surgery of genitalia within the first two years of life. Number of operations ranged from six to nine in these participants with hypospadias, including staged repairs and several reconstructions due to complications (range 6–40 years). Two male participants had Klinefelter syndrome (KS), one was diagnosed in early adulthood and the other in adulthood. Participants with KS reported no surgical interventions related to DSD.

All participants expressed identifying with the sex they were assigned at birth. The time towards the diagnosis of DSD was made, differed between and within diagnoses. Eleven participants were diagnosed before the age of 18, and four were diagnosed as adults from the age of 18 years to 35 years. See Table 1 for a brief description of the five diagnoses.

All participants had received medical follow-ups in childhood. None reported receiving a multidisciplinary follow-up as adults. All of the participants spoke Norwegian as their native language. Individuals with a condition defined as DSD, as described by Lee et al. (2006), were invited to participate. The exclusion criterion was the presence of an intellectual disability that affected the ability to participate in the interview process (no individuals were excluded from the study).

Procedure and measures

Clinicians representing the two multidisciplinary DSD teams in Norway contributed to identifying all eligible participants by searching for relevant diagnostic ICD-10 codes in their institutions' electronic health records. In addition, two national competence centers for rare disorders contributed to identifying eligible participants from their registry. Patient support groups and four organizations for Lesbian, Gay, Bisexual, Transgender and Intersex (LGBTI) working for equal rights for people who challenge the norm for gender and sexuality, promoted to participation by spreading information about the study on their webpages and social media channels. All of the potential participants received written information about the study and a consent form by mail. After we received their signed consent forms, the participants were contacted by telephone to arrange a time and place for the interviews.

Each participant was offered the choice between a face-to-face interview and a telephone interview. Nine participants chose a face-to-face interview, while six participants

Table 1. Brief description of the represented conditions.

Diagnosis	Brief description	References
Congenital adrenal hyperplasia (CAH)	CAH affects both males and females. Persons born with CAH lack an enzyme that the body needs to produce cortisole and aldosterone, two vital hormones. Consequently, the body produces more testosterone than needed. For girls, this may result in genital variations such as a larger than typical clitoris and a closed vaginal opening. Persons diagnosed with CAH are in need of lifelong medication to normalize their hormone levels	Witchel (2017)
Mayer-Rokitansky-Küster-Hauser syndrome (MRKH)	MRKH affects only females. The ovaries and external genitals are normal and females with MRKH develop breasts and pubic hair. However, females born with MRKH have a uterus, cervix and upper vagina that has not developed as expected. Consequently, they do not start to menstruate and cannot become pregnant. Penetrating intercourse might be difficult because of a shorter vagina	Herlin, Petersen, and Brännström (2020)
Turner syndrome (TS)	TS only affects females. They lack partly or completely the one X chromosome. The most common future of TS is short stature and non-function ovaries, resulting in a lack of monthly periods and infertility. TS is often associated with a number of other health conditions and symptoms, including learning difficulties and social problems	Shankar and Backeljauw (2018)
Klinefelter syndrome (KS)	KS only affects males. Individuals with KF are born with an extra X-chromosome (XXY) and do not produce the usual level of testosterone. Males with KS have differences in the development of male characteristics (testes and body hair), delayed puberty and KS may affect bone strength and fertility	Tremblay et al. (2016)
Hypospadia,	Hypospadia only affects males and affects the development of the penis. The types of hypospadias range from the urethral opening appearing nearer the tip of the penis or nearer the scrotum The testis may be affected	Kumar and Cherian (2022)

opted for a telephone interview. The face-to-face interviews were conducted at the Oslo University Hospital in Norway, and the relevant participants' travel expenses were reimbursed. All interviews were conducted from May to September 2018 by two female authors (A. W. and C. H.), a child psychiatrist who works in the field of DSD, and one with a master`s degree in psychology. The participants had never met the interviewers before. The interviews lasted between 45 and 90 min. The interviews were audio recorded using a Zoom H2n Handy Recorder and transcribed verbatim by CH and two research assistants. The participants were de-identified in the transcripts. The interview guide was designed to elicit accounts of the participants' experience of everyday life with DSD. Participants were asked open-ended questions covering a wide range of themes, including romantic relationships, experienced discrimination, satisfaction with surgical / medical / psychological treatment, information received since diagnosis, and disclosure. The participants were encouraged to describe their experiences, and follow-up questions were used to prompt the participants to elaborate on relevant issues or to offer examples to illuminate their stories (Kvale & Brinkmann, 2009).

The study formed part of a larger research project commissioned by the Norwegian Directorate for Children, Youth and Family Affairs (Feragen, Heggeli, & Wæhre, 2019) which aimed to explore the group's life situation and requirement for health and care services and interventions. The report is available in Norwegian with an

English abstract and has not previously been published in English. A total of 334 invitations were distributed, and 83 signed consent forms were received (aged 18-70). The response was higher in some groups (TS and KS). A purposive sample of 27 people was drawn to have participants representing age, gender, geographical affiliation and diagnosis.

In the present study, we analyzed and reinterpreted data concerning a subset of the original adult sample involved in the main project. The 15 participants in our study were selected on the basis of their age (30 years and older). The reason for this was three-fold: first, we wanted to restrict the age range from the original 18–70 years; second, there is an ongoing parallel sub-study examining the lived experiences of young adults; and third, we wanted to dive deeper into the data than what had been done in the initial analysis that was more descriptive.

In order to secure patient and public involvement, a reference group was established as part of the larger research project that comprised user participants, LGBTI activists, patient organizations, and professionals with legal, medical, and psychological backgrounds. The reference group represented a variety of gender perspectives and medical and legal interests.

Ethics

This study was conducted in accordance with the principles of the Declaration of Helsinki. All protocols and methods were approved by the Norwegian Regional Committee for Medical Research Ethics in South-Eastern Norway (number 79444) and by the Data Protection Officer at Oslo University Hospital (number 7000898). Due to the sensitive nature of the topics discussed during the interviews, the participants were offered a follow-up conversation after the interview. No one expressed a need for this.

Data analysis

The qualitative data obtained from the interview transcripts were first assessed independently (L. M. M.) and then collaboratively by four of the authors (A. W., L. F., L. M. M., and S. S.). These authors comprised a group with a variety of professional background (a nurse, a clinical psychologist, a child psychiatrist and a researcher in medical humanities), of which two are working within the DSD field. The data analysis was drawn on both Braun and Clarke's (2006) six-stage process and the principles of reflexive thematic analysis (Braun & Clarke, 2019). Reflexive thematic analysis is recognized as a suitable method for identifying patterns of meaning across datasets as well as divergence within data (e.g. between diagnosis, age at the time of diagnosis, and gender) (Braun & Clarke, 2019). Meaning requires interpretation, and it is not self-evident within data (Braun & Clarke, 2019). In this study, familiarization with the data was achieved through reading, re-reading, and making notes in the margins while striving to keep an open mind. The data were inductively coded (and recoded) by hand by the first author with the aim of identifying the participants' personal and pre-reflexive experiences of disclosure (Braun & Clarke, 2006). First, the codes focused on each participant's experiences as they appeared in the transcribed material. Next, the researchers searched for categories, similarities and divergences. In this process, we identified two themes.

Drawing on these themes, we read through the interviews once more to notice how these two strategies were expressed by the participants. To elucidate each theme, the researchers provided a selection of illustrative quotations, which were slightly revised to improve the readability, in accordance with the approach of Kvale and Brinkmann (2009). Quotes from the interviews were translated from the original language into English. In what follows, pseudonyms are used to protect the confidentiality of the participants, and diagnosis and age range are reported to increase readability.

Results

Participants represent a heterogeneous group of individuals affected by DSD. Two major themes were generated from the data regarding experiences of daily living: (a) Hiding a different body: A way of managing being different; and (b) Revealing information: From coerced exposing to acceptance of ambiguity.

Hiding a different body: a way of managing being different

All participants disclosed a story about a body that functioned or had an appearance that in some way was different from most other bodies. Two sub-themes were generated: (a) Concealing a functionally different body; and (b) How am I perceived by others?

Concealing a functionally different body

Being born with the condition that affects sex development affects how the body works in different ways. Most people take body functions for granted, e.g. like standing and peeing for men, using a tampon, being pregnant or being able to carry out sexual intercourse with penetration. For the participants, functionality seemed to take a big focus of attention. At the same time, the effort involved in concealing their difference was not necessarily visible to other people, and sometimes not even to themselves, which suggests that this effort may be understood to comprise internalized actions they were unaware of. Such actions included, for example, detailed planning, making up excuses, and avoiding situations such as dating, sport activities, and using communal showers.

The male participants described the importance of having a penis that was functionally 'normal' and how this affected their everyday life. Peter described how he 'took for granted' (his words) what he did to compensate for not being able to stand up and pee without spilling urine, and how this is actually not something 'normal' people do:

> This is why I like it when you can lock the door behind you at a public toilet, because then you can … (pauses), if you are peeing, you can clean up afterwards […]. Because it is not cool if your buddies come in after you and see the mess […]. You think about this when you are out on the town, when you are at home and need to use the bathroom, when you are going to have sex, when you are going to work, taking a bath, everything (hypospadias, 30–49 years).

Two thirds of the participants spoke of taking precautions in relation to dating and intimacy, and how they struggled with such issues, dedicating a lot of mental resources to planning when it came to concealing their difference. Christian, one of the male participants with KS, discussed how having a small penis affected his relationships with women

and how this resulted in avoidance behavior: 'I can talk to women … that's not the problem, but if it kind of advances to the next level, then I withdraw. It has to be really special before I take the next step' (KS, 50–70 years).

Most participants seemed unaware of the invisible work they engaged in. Laura, one of the women born with CAH, stated: 'In a way, there is nothing different about being born with this [CAH]. It's the same as being born with a missing arm, or a heart condition, or something. Only, the consequences of it [CAH] can, of course, be very different' (CAH, 30–49 years). When she talked about the consequences of CAH, she appeared to be referring to her virilized genitalia, which for many years caused her to believe that she was unable to have sex. For Lisa, the information she received about MRKH when being diagnosed (i.e. information about her vagina being too shallow and requiring vaginal dilatations), affected her sex life in a negative way. Before diagnosis she had an uncomplicated sex life, but afterwards it became difficult: 'I was single for many years and dreaded having sex […] and it actually made me avoid sexual contact with men for several years' (MRKH, 30–49 years).

How am I perceived by others?

Being born with a condition that affects sex development, not only affects the body's functionality but may also affect what the body looks like and how others perceive you. In the same way, as participants avoided situations where someone could discover the different functionality with their body, they were afraid of being perceived as different. This fear seemed to affect the balance between concealing and revealing personal information.

Having thoughts about gender identity, and which gender role you possess or fit into both in the gaze of others and in your own view, is a part of living with DSD for some participants. Susanna discussed how lack of information from clinicians and parents and communication about physiological and psychological processes affecting CAH caused her to worry about who she was and where she fitted in: 'You kind of felt like the identity-part was a bit difficult then. […] I am, well, what sex am I? In a way […]. Yes, it was probably during adolescence' (CAH, 50–70 years). Although all of the participants expressed that they had reached a point in life where they were confident about their gender identity, they continued to feel afraid of how they might be perceived if their peers knew about the DSD condition. This made them avoid situations such as dating, sports, and using communal showers. Christian, mentioned how traveling with colleagues left him in a difficult position when it involved spending the night in a hotel with colleagues: 'It was a nightmare if I had to share a room with one, two, or three others. We had to share showers and all that' (Christian, KS, 50–70 years). He explained how he came up with excuses and told lies to avoid sharing rooms so that no one could reveal his bodily difference. The use of phrases such as 'it was a nightmare' indicates that sharing rooms may be understood as something that both threatened the concealment of his condition and created a lot of effort and lies.

The invisible work to avoid being revealed having a DSD condition affected the childhood and youth in a significant way and continued to affect the daily life as adults. Peter, who was a talented athlete, chose to discontinue as an athlete due to his fear of his atypical penis being revealed: 'I avoided all sports, all team sports […]. I did not want to

shower. No, it was a big deal. That was mainly the thing' (hypospadias, 30–49 years). All of the male participants reported taking precautions when faced with having to use a communal shower as an adult, such as checking the locker room for separate showers or going home to shower after sports. The women on the other hand, seldom talked about public appearance, but more about how it affected intimate and private situations.

Most participants seemed unaware of the invisible work they engaged in, giving contradictory narratives. For instance, one male participant indicated that no partner had ever commented on the appearance of his penis and that he was content with the look of it. However, he expressed that he was unlikely to date girls in case they found out about his unusual genitalia.

Revealing information: from coerced exposing to acceptance of ambiguity

The decision to reveal information about DSD consisted of dilemmas and a balancing act between a need to control what others might think of them and a need to tell about it. Three different subthemes were generated that influenced disclosure: (a) the context; (b) time of diagnosis; and (c) whether they mastered an everyday language.

To disclose, or not to disclose? Context matters

In this subtheme, participants reveal how they prefer to conceal personal information. Yet, in order to have intimate or close relations, they may see the need for disclosure or feel that it is necessary. Issues like fertility, an altered appearance of genitalia or difficulties with having sex were important issues in this regard and shared by most participants. Laura experienced a strained relationship to sex all her life. When she met a partner she became serious with, she felt obligated to reveal sensitive information about the parts of her body that were private and different:

> Well, it was necessary when sex became an issue. I knew she would understand that something was a bit different […]. I felt a strong need to explain everything, about the operation and all those things […]. I didn't want them to think … , or to get strange fantasies or anything. I rather they knew (CAH, 30–49 years).

Some of the participants expressed how disclosure was considered a positive thing, when the recipient of the disclosure had some knowledge and/or interest in the matter. Sebastian experienced that it was easier to talk to others living with illnesses. 'The only one I can talk to is my father … because he also has a.. not a syndrome.. but a (diagnosis), so I can talk to him, […] he understands' (KS, 30–49 years).

For most participants born with KS, TS and MRKH, infertility was an issue. The balance between feeling responsible for informing their partner and feeling a need to conceal infertility was a dilemma. It influenced intimacy, romantic relationships and contact with friends and acquaintances. This could involve avoiding social events and situations where they expected questions regarding, e.g. pregnancy, but for a few, it meant an opportunity to speak about infertility hoping to normalize and reduce the stigma that surrounded not having children. Ella's words describe the issue of infertility:

> So I think it's kind of OK to tell about (infertility), because it's a way to make sure that people don't run after me and ask when I am going to have children all the time, because

> that's very tiring. […] One of the few things I avoid in everyday life is baby-shower and stuff like that. It can get a little tough (MRKH, 40–50 years).

Disclosure as a time-dependent phenomenon

In spite of the heterogeneity of DSD, a dichotomy appeared through the participants' experiences: those who learned about their condition during childhood or whether they had been diagnosed as adolescents or adults (during/after puberty).

Several participants who were diagnosed during childhood ($n = 11$) commented that as they grew older, they became more at ease with their body and diagnosis. This resulted in them reaching a level of acceptance of their differentness, which made revealing information about their bodily differences easier. As Thomas expressed: 'Well, I think I would have had more issues with talking about it 15–20 years ago. I wasn't as open as I am now' (hypospadias, 30–49 years).

Four participants who were diagnosed with DSD later in life felt that disclosure became more difficult after being informed about their condition. Ingrid learned about the condition when she was an adult and had not been aware that she might be perceived as 'different'. She started reading about TS: 'And, I was shocked. […] Abnormal breasts, and private parts, and … So I thought, thank god I was married, otherwise I wouldn't had the guts to get involved with a man' (TS, 50–70 years). Participants with KS described how the diagnosis generated an awareness of their bodily differences. It became problematic to reveal their bodies at the beach, at the gym, in dressing rooms, and in intimate situations.

> I got a slap in the face then. So yes, I still struggle with it a bit, mentally, I actually do […]. When I was younger, I didn't care, but now I have learned about [the consequences of KS], I struggle to take off the clothes on my upper body […]. I feel that sex might be a bit more difficult now. Also, because I've figured this out, I've realized that my testicles should be much larger (Sebastian, KS, 30–49 years).

Mastering an everyday language

Participants expressed how they did not have a way of talking about DSD so that others could understand. Some even expressed how they lacked knowledge about the condition and how this affected how they talked about it. Jane for instance, explains:

> If they ask, I tell them that my body produces more testosterone than your body does, and that I need to take medications to balance this. Then they reply: «oh, ok». Because I … I cannot give them any more information, because I don't know any more (laughs) (CAH, 30–49 years).

Some participants mentioned how their lives would have differed in a positive way if someone had taught them how to use an everyday language to communicate their DSD:

> I didn't tell anyone about it, didn't communicate anything about hypospadias to anyone until I was 20, 19, maybe 18 years old. It was a big secret in a way. It just went like that. I think this is the reason why it's important to encourage children to talk about it, because otherwise the problems will escalate, rather than you understanding that it is really not such a big deal (Peter, hypospadias, 30–49 years).

Participants may thus realize that gaining knowledge of DSD and having the ability to communicate about their condition might be primarily positive for their own understanding and well-being.

Discussion

The main finding of this study concerned how adults with DSD struggled with reaching a balance between information sharing and concealment. The avoidance behaviors exhibited by several of the participants may imply that they anticipate stigmatization or that stigma was internalized. Stigma may be generated by what is defined as undesirable and discrediting attributes that people typically seek to avoid. Fear of stigmatization could possibly explain the ambivalent component of disclosure; several commented on disclosure as important, that children and youth should be trained in practicing disclosure, while they, in contrast, did not feel comfortable with revealing private information.

Impact on everyday life

Challenges concerning the disclosure of information about their bodily difference are central to everyday life and constitute an important facet of the experience of living with DSD. Previous research has stated that children and adolescents living with DSD born before the consensus statement of 2006 have experienced too little information and inadequate communication about DSD (Howe, 2021; Lee et al., 2016). Based on this knowledge, it is not surprising that our participants struggled with disclosure. However, it is important to note that none of the participants had received a multidisciplinary follow-up as adults. They had all reached adulthood in 2006, an age group for whom no routine follow-up has been implemented so far. This may be one of the reasons why they continued to struggle in silence or used mental resources to decide whether to hide or reveal what they perceived as different.

A quantitative study including 1040 participants born with DSD found that a positive overall body image was associated with disclosure about one's condition (van de Grift, Cohen-Kettenis, de Vries, & Kreukels, 2018). In our study, participants had an ambivalent relationship with disclosing information, even if disclosure usually was perceived as a positive experience. Disclosure concerning their different body was reported as unnecessary, except in situations where it would be visible. This ambivalence has also been described in other studies (Lampalzer et al., 2021; Sharratt, Williamson, Zucchelli, & Kiff, 2020) and may be related to feelings of stigma. Similar observations are documented in other populations affected by chronic and/or congenital conditions. The stigma surrounding HIV/AIDS is well known. Other conditions that affect intimate parts of the body, e.g. fecal incontinence, could also be comparable, as it represents a taboo with bodily functions that will only be discernable in certain situations (Chelvanayagam, 2014). Chelvanayagam (2014) describes how people with gastrointestinal conditions who are hyper-vigilant for signs of possible social rejection or discretization, may try to conceal the difference if possible, by using defensive or avoidance strategies. Perceived stigma related to the pressure to pass as able-bodied was also reported in a large qualitative analysis following an online survey including

a whole range of different rare conditions (Munro, Cook, & Bogart, 2021) and has also been discussed in relation to visible conditions (Germain et al., 2021; Masnari et al., 2013). People with DSD and other chronic conditions with feelings of internalized stigma are less likely to discuss the taboo openly, may not possess the appropriate vocabulary, and may fear that healthcare providers treat them with prejudice and discrimination (Earnshaw & Quinn, 2012). Hence, feeling of stigma can result in a reluctance to access care.

The lack of understanding from friends, families and health personnel can increase feelings of loneliness and stigma. The limited knowledge among people in general about DSD, as well as their lack of knowledge about how genetic, gonadal, and hormonal factors can affect individuals with such conditions, may add to the burden faced by individuals with DSD. This should be taken under consideration when planning on how and what to tell others. In a qualitative study, Engberg et al. (2016) described how individuals with DSD considered their condition as too complex to explain to others. Research shows that this may result in communication difficulties with health professionals indicating the need to develop vocabulary that can be adapted to different situations (Sanders & Carter, 2015). Our findings indicate that patients, parents, and clinicians lacked an everyday language for talking about differences in bodies in general as well as the impacts of bodily differences on psychosocial aspects of daily life and body image in particular.

Invisible work

The participants in the present study, who lacked the opportunity to talk about their diagnosis while growing up, later engaged in invisible work to achieve a balance between concealing and revealing their difference. Furthermore, this invisible work seemed to be an effort to remain in control of the information flow and to avoid being 'revealed' (i.e. anticipated stigma). In particular, experiences with an altered genital appearance or function caused difficulties in terms of sharing information about their bodily deviations because they did not have sufficient knowledge about their differences. This finding is in line with several other studies (Alderson, Madill, & Balen, 2004; Engberg et al., 2016; MacKenzie, Huntington, & Gilmour, 2009; Meyer-Bahlburg et al., 2017). For our female participants with TS, their invisible work seemed to be focused on short stature, social problems, and infertility, as shown within the TS literature (Nisbet, 2020; Sutton et al., 2006).

Participants' accounts of doing preparations before revealing information about the condition is not unique to DSD. In a study done by Sharratt et al. (2020), participants with different visible but concealable conditions (e.g. skin conditions or burn scarring on parts of the body that may not necessary be visible), discussed different ways and situations in which they took control over the disclosure process, e.g. by selecting the timing, location, and level of disclosure. Preparing disclosure was a way of controlling what others knew about their condition and worked as a coping mechanism, as illustrated in the present study.

Conducting in-depth interviews appeared to be important in terms of reaching a better understanding of the complex relation between disclosure, stigma, and everyday

life. The interview guide did not contain questions that explicitly focused on 'stigma'. However, the participants frequently reported narratives of shame, differentness, and coping mechanisms, indicating that participants did not have an awareness of the invisible job they did.

Time of diagnosis

A diagnosis received early or later in life revealed a discrepancy across two groups. As stated above, growing older had a positive impact on feelings of difference in participants diagnosed in childhood, leading to acceptance of their own identity and, in some situations, a reduction in invisible work. Interestingly, the greatest discrepancies between the diagnostic groups were for participants diagnosed as adults. The information they received about having DSD made talking to others, getting undressed in public, and participating in intimate relationships more difficult than prior to receiving a diagnosis. This was the case for males with KS and females with MRKH. Guntram (2013) investigated how women found out about their atypical sex development during adolescence and how they considered themselves as either 'normally different' or 'differently normal'. This reflected how they understood the diagnosis; a source of stigmatization and medicalization, or a way to make sense of their new situation. A medicalized language may alienate the patient from the condition and make him/her feel that the body is somehow diseased and something that needs to be fixed, something others might have problems to accept. MacKenzie et al. (2009) suggested that people may develop acceptance of their differences when they learn about it, have someone to talk to, and receive support from family and friends. This suggests that the dissemination of information needs to be sensitive and customized, even when the patient is an adult. Our results also indicate that healthcare professionals should examine how new information is perceived and understood, and how it potentially influences the affected persons' identity and psychological well-being.

Strengths and limitations

Few studies have explicitly focused on how adults over the age of 30 experience living with DSD. The result derived from the present study could therefore be useful for clinicians and researchers to understand how it is being an adult with DSD, and how we can better help those who need it. The study has a qualitative approach, and a relatively heterogenic population. The diversity of diagnoses might limit the generalizability of the findings. However, the explorative and qualitative nature of the study made it possible to generate themes across the material but also deviations within the material and gave us rich and nuanced examples suitable to illuminate the experienced phenomena from the participants' own perspectives. In this study, the interviews were conducted in 2018. To our knowledge, health care follow-up has not changed during the last four years or during the COVID pandemic. Therefore, we considered that current findings do not demonstrate any time-related impact on how participants may have experienced health care follow-ups differently compared to if the interviews were conducted today.

The participants in this study were all born before the 2006 consensus statement and since then there have been changes in psychological health care and follow-up. Thus, the

present findings should not be generalized to younger age groups because children born with DSD after 2006 might have received a different multidisciplinary follow-up.

Despite involving LGBTI organizations in the recruitment, we did not receive any participants from these channels. In addition, during the recruitment phase, predominantly people with KS and TS expressed an interest in participating. To achieve balance in terms of diagnostic representation, a purposive sample was chosen, in which the spread with regard to age and geographical location was taken into account. Yet, the sample might have resulted in a selection bias as we had no background information about the severity of their condition or other potentially important factors when recruiting. The reference group was used to check for relevance, and they gave positive feedback on results being recognizable and relevant to their daily life. Finally, all of the participants were Caucasian. Future studies should aim to recruit individuals from different ethnic and racial backgrounds.

Conclusion

It is important that people with DSD receive appropriate information and has someone to talk openly to about the psychosocial aspects of living with their condition. Individuals with DSD need lifelong multidisciplinary follow-up and renewed information that is adjusted to the timing of diagnosis, life situation, and psychological status. This requires sensitivity and pacing of information sharing from both families and clinicians, and the necessary information should be provided in an individually adapted and personalized manner.

Disclosure and communication about DSD during adulthood can create or enhance inherent feelings of distress, stigma and lack of belongingness. The participants' anticipation of reactions to their differentness seemed to have a major impact on what they chose to share, even as adults. Silence may lead to both anticipated and internalized stigma and increase the suffering in individuals with DSD. By increasing awareness and reducing misconceptions in the community, clinicians can influence the feeling of being accepted. We also need to recognize the impact on everyday life of working to hide or choosing to disclose a body affected by DSD. Awareness about differences such as DSD need to be dealt with at an interpersonal level, a community level, and last, but not least, on an institutional level so that people with DSD may avoid being exposed to attitudes representing outdated knowledge towards individuals with diversity in sex development.

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Data availability statement

Due to the qualitative nature of this research, the datasets generated during the current study are not publicly available due to participant confidentiality issues.

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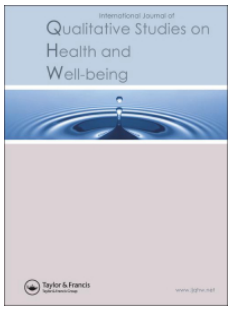

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Understanding sexual health concerns among adolescents and young adults with differences of sex development: a qualitative study

Line Merete Mediå, Solrun Sigurdardottir, Lena Fauske & Anne Waehre

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EMPIRICAL STUDIES

Understanding sexual health concerns among adolescents and young adults with differences of sex development: a qualitative study

Line Merete Mediå [a,b], Solrun Sigurdardottir [a], Lena Fauske [b,c] and Anne Waehre [d,e,f]

[a]Women and Children's Division, Centre for Rare Disorders, Oslo University Hospital, Oslo, Norway; [b]Department of Interdisciplinary Health Sciences, Institute of Health and Society, University of Oslo, Oslo, Norway; [c]Department of Oncology, Norwegian Radium Hospital, Oslo University Hospital, Oslo, Norway; [d]Department of Child and Adolescent Psychiatry, Oslo University Hospital and Institute of Clinical Medicine, Oslo, Norway; [e]Division of Endocrinology, Boston Children's Hospital, Boston, MA, USA; [f]Department of Pediatrics, Harvard Medical School, Boston, MA, USA

ABSTRACT

Purpose: Differences of sex development (DSD) are congenital conditions that involve variations in individuals' sex chromosomes, genes, external and/or internal genitalia, hormones, and/or secondary sex characteristics. This study sought to elucidate the experiences of adolescents and young adults living with DSD by focusing on their experiences of intimacy and sexual health.

Methods: An interpretative phenomenological research design was adopted. Semi-structured qualitative interviews were conducted with 11 Norwegian adolescents and young adults aged 16–26 years who had five different DSD conditions. The interview findings were analysed by means of a reflexive thematic analysis.

Results: The participants reported feeling different, both in terms of how their body functioned and how their body looked. Lack of knowledge increased this feeling of differentness. Moreover, lack of everyday language with which to talk about intimacy and sexual concerns resulted in the participants feeling stigma. Anticipating stigmatization and lacking everyday language complicated the participants' communication regarding their DSD and sexual health.

Conclusions: The sexual experiences of adolescents and young adults with DSD are diverse. Fear of stigmatization and lack of everyday language complicate communication with healthcare professionals and others. Understanding their unique needs is crucial to helping individuals achieve good sexual health.

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Disorders of sex development; differences of sex development; intersex; sexual health; stigma; communication

Introduction

Differences of sex development (DSD) represent a heterogeneous group of congenital conditions that cause the development of the genitals, hormones, and/or chromosomes to differ from traditional conceptions of male and female bodies (Lee et al., 2016). As a group, DSD have an estimated incidence of 1:4500 births (Sax, 2002). A DSD may become evident at birth or during childhood, adolescence, or adulthood, depending on the specific condition and its severity. Different DSD aetiologies, treatments, and individual experiences may affect individuals' sexual health (Amaral et al., 2015; Wisniewski et al., 2019). In fact, adolescents and adults with DSD have highlighted sexual aversion and lack of arousal as the most common problems experienced (Hughes et al., 2006). WHO defines sexual health as "a state of physical, emotional, mental and social well-being in relation to sexuality" (World Health Organization [WHO], 2015, p. 5). The WHO definition is quite broad. More specifically, sexual health may also concerns gender identity, sexual orientation, eroticism, pleasure, intimacy, and reproduction (Graugaard, 2017). In 2006, a consensus statement was published in an effort to improve the management of individuals with DSD (Hughes et al., 2006). The statement offered suggestions for improving individuals' sexual health, including a focus on interpersonal relationships, referral to sex therapy, avoidance of unnecessary medical photography and/or genital examination, access to mental health professionals, and assessment of sexual health (Hughes et al., 2006). An update to the statement was published in 2016, which highlighted the role of psychoeducation in reducing anxiety related to sexual and romantic relations (Lee et al., 2016).

The body of literature concerning the sexual health of individuals with DSD has grown in recent years, although it remains both limited and characterized by contradictory results (Wisniewski et al., 2019). Some quantitative studies involving individuals with different DSD reported the participants to exhibit overall satisfaction with their sexual health (Engberg et al., 2022; Schönbucher et al.,

CONTACT Line Merete Mediå lmedia@ous-hf.no Centre for rare disorders, Oslo University Hospital (Rikshospitalet), Postboks 4950 Nydalen, Oslo 0424, Norway

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2008), whereas other quantitative studies found that DSD had a negative impact on participants' emotional and sexual well-being (Liao et al., 2011 van de Grift et al., 2022). A meta-ethnography of 16 qualitative studies concerning DSD suggested sexual health to be negatively related to DSD and, consequently, quality of life (Sani et al., 2019). The medical consequences of DSD (type and severity), psychological experiences of treatment (e.g., distress, anxiety), and reactions from others (e.g., stigma) have all been found to have a negative influence on sexual health (Meyer-Bahlburg et al., 2018; Wisniewski et al., 2019). Furthermore, the surgical management of DSD has been determined to be associated with both positive and negative consequences with regard to individuals' sexual health (Sani et al., 2019).

Over the past decade, there has been an increased focus on the relation between sexual health and both physical and emotional health (Graugaard, 2017; Rew, 2006; WHO, 2015). It has been established that adolescents and young adults (AYA) with a positive perception of their body and good knowledge of sexuality often exhibit better physical and psychological health when compared with AYA who lack knowledge and personal control over what happens to their body (Callens et al., 2021; Rew, 2006). However, only a limited number of studies have investigated sexual health among an AYA population with DSD. Thus, the present study sought to increase the understanding of the experiences of AYA with DSD by focusing on their experiences of intimacy and sexual health in a broad sense, and let the data and the analytical process guide the focus of this study.

Methods

Design

We sought to understand the lived experiences of AYA with DSD and so applied an explorative qualitative research design that was epistemologically grounded in the hermeneutic phenomenological tradition (Kvale & Brinkmann, 2009). Hermeneutic phenomenology focuses on the first-person perspective as experienced by the individual themselves as well as on how the researcher interprets meaning based on their professional knowledge (Giacomini, 2010). More specifically, the interpretation is based on both the participant's and the researcher's preunderstandings, as well as on the research context, and it develops throughout the entire research process (Giacomini, 2010). To capture the experiences of AYA living with DSD, interviews were conducted to collect data. In addition, to ensure affected individuals and public involvement, a reference group of AYA (three females and one male) with personal experiences of living with four different DSD was established.

Recruitment and participants

This study was conducted in Norway. The recruitment period ran from August 2020 to June 2021, and the study included two out of three subgroups of DSD: 46,XY DSD and 46,XX DSD. The inclusion of DSD conditions was based on the classification from Hughes et al. (2006) and Cools et al. (2018), including e.g., Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) and proximal hypospadias. The exclusion criteria were AYA with sex chromosome DSD. We chose to include different DSD conditions and all genders in order to elucidate the similarities and differences between the diagnoses among AYA with DSD aged 16–26 years.

We aimed to capture AYA with verified diagnosis of DSD by recruiting participants from the two multidisciplinary DSD teams based at Oslo University Hospital and Haukeland University Hospital and the Center for Rare Disorders at the Oslo University Hospital, Norway. Consequently, we did not aim to include individuals with DSD from a community sample and the perspectives of human rights defenders or activists were not explored in this study.

Thirteen individuals signed the consent form and were subsequently contacted by telephone to arrange a time and place for the interview. One individual had a sex chromosome diagnosis and was excluded, whereas one individual did not attend the scheduled interview. The recruitment size was smaller than expected and the enrollment was time consuming due to several issues. The researchers did not approach AYA directly and had to rely on clinicians for recruitment. In addition, participants were recruited during the COVID-19 pandemic and AYA might thus have cancelled the clinical follow-ups.

The information gathered about diagnoses and treatments was based on the participants' own reporting. The participants reported five different conditions: congenital adrenal hyperplasia (CAH), proximal hypospadias and/or complex syndromic structural associations of male genital development, Mayer-Rokitansky-Küster-Hauser syndrome (MRKH), Swyer syndrome, and complete androgen insensitivity syndrome (CAIS). Table 1 presents brief descriptions of the five conditions.

The 11 participants differed in terms of their gender, age (range: 16–20 years [$n = 5$] and 21–26 years [$n = 6$]; mean age: 20.3 years), time of diagnosis (at birth/infancy [$n = 6$], puberty/early adolescence [$n = 5$], and number of surgeries. Most participants had an ethnicity originated in the country where the study was conducted. All participants identified with

Table I. Brief descriptions of the represented conditions.

Diagnosis	Brief description	Reference
Congenital adrenal hyperplasia (CAH)	CAH affects both males and females. Persons born with CAH lack an enzyme that the body needs to produce cortisol and aldosterone, two vital hormones. Consequently, the body produces more testosterone than required. For females, this may result in genital variations, such as larger than typical clitoris and a closed vaginal opening. Persons diagnosed with CAH require lifelong medication to stabilize their hormone levels.	Witchel (2017)
Mayer-Rokitansky-Küster-Hauser syndrome (MRKH)	MRKH only affects females. The ovaries and external genitals are normal, and females with MRKH develop breasts and pubic hair. However, females born with MRKH have a uterus, cervix, and upper vagina that has not developed as expected. Consequently, they do not start to menstruate and cannot become pregnant. Penetrating intercourse might be difficult due to a shorter vagina.	Herlin et al. (2020)
Swyer syndrome	Females with Swyer syndrome have a female phenotype. They have a uterus, vagina, and fallopian tubes. The gonads have not developed as expected and produce no hormones. They have a 46,XY karyotype and do not menstruate. Females with Swyer syndrome cannot have genetic children, although pregnancy may be feasible through egg donation. Early prophylactic removal of the streak gonads is discussed due to the risk of developing gonadal malignancy. Hormonal therapy is required.	Michala and Creighton (2010)
Complete androgen insensitivity syndrome (CAIS)	Individuals with CAIS exhibit hormonal resistance to androgens, which results in a female phenotype with a 46,XY karyotype and testes that produce androgens (testosterone). Consequently, females with CAIS have an absent uterus, shorter vagina, are infertile, and do not menstruate. Prophylactic removal of the gonads (testis) is considered due to the risk of developing gonadal malignancy. Hormonal replacement therapy is offered.	Grymowicz et al. (2021)
Hypospadia, severe form	Hypospadia only affects males. It affects the development of the penis. The types of hypospadias range from the urethral opening appearing nearer the tip of the penis or nearer the scrotum. The testis may be affected.	Kumar and Cherian (2022)

the assigned sex at birth (male or female), no one identified themselves as non-binary. Table 2 presents participants' gender, age and diagnosis. Six of the female participants had 46,XX DSD. Of these, three females were diagnosed before the age of two years. The other three were diagnosed later in childhood and adolescence (age range: 7–16 years). Five of the 46,XX females reported having undergone a primary surgery to the genitalia or removal of the inner genital structures (either before two years of age, at a prepubertal age, or during puberty). Three had undergone additional genital surgeries later in childhood or adolescence, and two had plans for further surgeries in the near future. In addition, five participants took glucocorticoid replacement therapy.

The 46,XY female group comprised two females with different diagnoses. They were both diagnosed in adolescence and had undergone surgical removal of gonads (gonadectomy), uterine rests, vaginal dilations, and/or surgical incision. Both 46,XY females had received information about the use of vaginal dilators. In addition, they both took sex steroid replacement therapy.

The three male participants all had proximal hypospadias and/or complex syndromic structural associations of male genital development. Furthermore, they had associated congenital anomalies (e.g., skeletal, kidney, gastrointestinal tract) and were diagnosed within the first two years of life. They had undergone surgeries to the genitalia within the first two years of life. One male had undergone three surgeries in adolescence, while another was waiting for reconstructive genital surgery. The third male reported needing a hypospadias correction. The male participants are referred to as XY males in this study.

Seven participants had sexual experiences with a partner and eight participants were or had been in romantic relationships. Most participants reported that they had been in love. Seven participants reported sexual attraction to a person of the opposite sex. Two of them had also thought about sexual relations with a person of the same sex, although they identified themselves as heterosexual. Three participants reported sexual attraction to the same sex, whereas one participant reported not yet knowing to whom the person was attracted to.

All the participants had received medical follow-ups prior to turning 18 years old. None of them had received a multidisciplinary follow-up after the age of 18 years. Three participants had received psychological treatment as adults. All participants diagnosed in childhood

Table II. Sample characteristics.

		Total ($n = 11$)	Male ($n = 3$)	Female ($n = 8$)
Age range (mean)		16–26 (20.4)	16–23 (20.6)	16–26 (20.3)
46,XX DSD female	CAH (n)	5	-	5
	MRKH (n)	1	-	1
46,XY DSD female	CAIS (n)	1	-	1
	Swyer syndrome (n)	1	-	1
46,XY DSD male	Hypospadia or structural associations of external genitalia (n)	3	3	-

Note: DSD = differences of sex development, CAH = congenital adrenal hypoplasia, MRKH = Mayer-Rokitansky-Küster-Hauser syndrome, CAIS = complete androgen insensitivity syndrome.

had received follow-up from a multidisciplinary team including a child and adolescent psychiatrist until the age of 18. Few participants talked about psychological support from a sexologist, endocrinologist or gynaecologist during adulthood. Participants diagnosed after 18 years old had not received psychological support on a regular basis from clinicians.

Data collection

The semi-structured interviews were conducted by the first author between October 2020 and June 2021. The dates and locations for the interviews were decided in collaboration with the participants. Due to restrictions necessitated by the COVID-19 pandemic, five interviews took place via video conferencing and one interview by telephone. Moreover, five interviews were conducted on a face-to-face basis, either in the participant's home ($n = 2$) or at University Hospital ($n = 3$). The interviews lasted between 35 and 123 minutes (mean: 63 minutes). All the interviews were audio recorded using a Zoom H2n Handy Recorder and then transcribed verbatim by the first author and a research assistant. The participants were de-identified in the interview transcripts.

The questions used in the interview guide were discussed by the research team and clarified in meetings with the reference group to ensure that topics were relevant and comprehensible (Tracy, 2010). The participants were asked open-ended questions covering a wide range of themes, including romantic relationships, satisfaction with surgical/medical/psychological treatment, information received since diagnosis, and disclosure. The participants were encouraged to describe their experiences in their own words, and follow-up questions were used to prompt them to elaborate on relevant issues or to offer examples to illuminate their stories (Kvale & Brinkmann, 2009). After each interview, the participants could make additional comments or ask questions to ensure their experiences of the relevant theme were understood. In addition, the first author wrote a short reflexive summary with the aim of capturing impressions not easily captured on audio recordings, such as changes in moods, body language, and facial expression.

Data analysis

The data were analysed in accordance with the principles of reflexive thematic analysis, as described by Braun and Clarke (2006, 2019). This method was chosen because a reflexive thematic analysis is recognized as a suitable method for identifying patterns of meaning across datasets as well as divergence within data (e.g., between diagnoses, age at the time of diagnosis, gender) (Braun & Clarke, 2019). To ensure the credibility of the findings, the authors double-coded a subset of interviews (50%) (Levitt et al., 2018). They began by familiarizing themselves with the data through reading and listening to each interview several times in order to gain an impression of the participants' experiences. The first author coded (and recoded) each interview inductively by hand on the basis of the participants' own words. Consequently, codes regarding sexual health were generated. Next, the researchers searched for categories, similarities, and divergences in the data. This step involved questions focusing on what the participants were telling about their sexual health, what was important to them, and how they were describing issues with intimacy and fertility.

Finally, all the authors agreed to the codes and themes that represented the findings. Engaging professionals with a variety of background in the research group was important to analyse and interpret themes from different perspectives and to ensure that the process was not misinterpreted (Tracy, 2010). This helped us to view the transcripts, interpretations, and results in different ways, to question each other's preunderstandings, and to gain a more in-depth understanding of the phenomena under study, thereby strengthening confirmability (Shenton, 2004). Throughout the entire process of analysis, the researchers regularly returned to the original data to check the themes and quotes and ensure that the meanings had not been lost during either interpretation or translation. The questions regarding sexual health were open-ended with the aim to let the participants guide the direction of this sensitive topic.

The reflexive summaries written after the interviews also formed part of the analysis. To elucidate each theme, the researchers identified a selection of illustrative quotes, which were slightly revised to improve their readability, as suggested by Kvale and Brinkmann (2009). All the quotes from the interviews were translated from Norwegian into English by the first author. To ensure rigour of the study the Consolidated Criteria for Reporting Qualitative Studies (COREQ) was followed (Tong et al., 2007), the checklist is available as supplementary material.

Ethical considerations

This study was conducted in accordance with the principles of the Declaration of Helsinki. All the protocols and methods were approved by the Norwegian Regional Committee for Medical Research Ethics in South-Eastern Norway (approval number 79,444) as well as by the Data Protection Officer at Oslo University Hospital (approval number 7,000,898). Due to the sensitive nature of the topics discussed, each participant was offered a follow-up conversation after their interview. Two participants wanted a follow up

conversation, and a referral to eligible health care personnel was arranged for.

When writing the results of this study, an arbitrary name was assigned to each participant to ensure their confidentiality. Moreover, the participants' age was dichotomized into groups of 16–20 years and 21–26 years for the purpose of de-identification. The same approach was adopted in relation to their medical conditions, which were organized into three groups: XX, females, XY,females, and XY,males.

Results

Themes regarding sexual health

Three themes regarding sexual health described the experiences of the participating AYA with DSD, namely the importance of being "normal," communication, and prospects of fertility.

The importance of being "normal"

The analysis generated subthemes wherein normalcy became relevant and described the effect on sexual health of "a body that is functionally different" and "a body with genitals that look different."

Two-thirds of the participants reported having had sexual experiences with a partner. They described sex and, in particular, intimacy as problematic to the extent of affecting their everyday lives. The female participants talked more about this topic than the male participants. The participants described how being functionally different influenced their sex life, for example, requiring preparation such as dilating the vagina, experiencing sex as painful, or feeling the need for genital surgery. Nora had difficulty with intercourse due to having a short and narrow vagina. She had wondered whether it would be better to date girls than boys: "I wasn't really able to have sex. Maybe I decided, that… Maybe I should like girls instead, because I struggled so much with dilating" (XY,female, 21–26 years).

Furthermore, two-thirds of the participants had undergone both invasive and non-invasive genital procedures. The operations involved constructive or reconstructive feminizing and masculinizing surgeries of the genitalia and/or urethra, or the construction of a neovaginal opening. Self-dilation therapy was often necessary for the maintenance of a functional vagina. In this regard, Ellida explained that vaginal dilation prevented her from being impulsive when it came to meeting a partner and having sex: "Yes, it [sex] is kind of problematic, because … I need to kind of do preparations like vaginal dilatation such a long time in advance… In a way, it kind of ruins the whole experience" (XX,female, 16–20 years).

For the female participants, experiencing their first menstruation, or the lack thereof, was described as characterizing an especially vulnerable period of life and representing a situation where their bodily function differed from that of their peers in a concrete way. Several participants explained that not starting menstruating at the same time as their peers was something they had learned to live with and found less problematic than what they felt clinicians expected them to feel. Agnes started her period earlier than her peers. Her doctor put her on hormones to postpone puberty, which resulted in side effects such as weight gain. Agnes related the following:

> I could easily have learned to live with it [menstruation]. Instead of just pushing it away and putting me on that hormonal shot […] It would have been better if someone could just have told me why I got my period early and explained how I could live with it. (XX,female, 16–20 years)

One of the female participants who had not started menstruating due to having a hormonal imbalance also did not problematize the issue: "I don't need to have menstruation, because I'm not going to have children anyway" (Rebekka, XX,female, 16–20 years). However, Nora, a young woman who required a medical examination due to amenorrhoea, talked about feeling abnormal: "Then I started wondering why … why everybody had started their period, except me … We used to talk about that person who got it so late and I started thinking, "Oh my god, am I that person?" (XY,female, 21–26 years).

In addition to having genitals that required "improvements" to be like those of others and function like they were supposed to, as well as having a body that did not menstruate like it should, the participants' sexual debut was an experience that seemed to significantly rely on society's norms regarding how to "be" a sexual partner. The participants did not seem to describe their first sexual experience as being related to their own pleasure and satisfaction. Rather, the question of whether or not it was considered a good experience appeared to be dependent on their partner's reactions. Nora had difficulty with dilation and did not have sex until she had undergone dilation in narcosis. When asked about how her first sexual experience went, she responded: "It went well … it did … we met two to three months after the surgery… it wasn't, kind of … he didn't notice anything, so it wasn't a problem" (XY,female, 21–26 years). Another young women described her sexual debut in the following way:

> The first time anyone was going to see me naked … Like in a sexual context, then… Well, it was really in

> the dark and it all went very well. He didn't sense or feel anything different about me […]. It was actually a good first time. (Agnes, XX,female, 16–20 years)

The normal appearance of the genitalia was important for some participants (CAH, XY,males). Having genitals that looked more "normal" was something all these participants wanted to achieve through surgery. Indeed, some were willing to pay for it if necessary (such surgery is normally covered by public healthcare in Norway). "That sucked, because I would like to have nice looking genitals. Everybody wants that. And it means a lot to me as an adolescent, not having a regular partner. […] I hope that I can get the surgery I want" (Agnes, XX,female, 16–20 years). Sex was described as problematic by the male and female participants who reported having genitals that were visibly different from the norm, which resulted in them expending a lot of mental resources on feeling different. Despite this, several participants described that when they had intercourse, it was unproblematic. They explained that this was because of the partner's response, which wasn`t described as negative, as mentioned above.

Some participants did not differentiate between appearance and function. Instead, they talked about the two as being dependent on each other, with appearance being more important than function. Agnes experienced reduced clitoral sensation. She was not pleased with the appearance of her genital and had recently undergone corrective surgery. She explained: "Because if the appearance had been, like perfect, I don't think I would have had such an issue with the functionality" (XX,female, 16–20 years).

Communication

Communicating about sexual health became an issue when the participants "lacked knowledge about their bodily differences and/or "lacked everyday language."

Not knowing why their body was different led to feelings of insecurity that affected the participants' sexual relationships. Thomas expressed having little knowledge about his DSD. He was unsure whether he had actually had an operation during childhood. He wanted to talk with doctors about the appearance of his penis but felt embarrassed about addressing the issue. He felt insecure in intimate relationships and avoided answering questions from partners: "Because I wasn't sure why [my penis looked different], I didn't have any answers as to why it looked different. […] So I didn't say anything. […] I kind of didn't know what to say" (XY,male, 21–26 years). By contrast, Henrik reported having regular discussions with his doctor and his mother about his DSD. Even though he acknowledged that adolescence was a particularly vulnerable period for a male whose penis looked different from the norm, he had the following to say about being intimate with a girl: "The more times you face it, the more confident you become that … You know what? It is not that important at all really" (XY, male, 21–26 years).

A difference was observed among the participants in terms of how they expressed their experiences with sexual intimacy. Several reported lacking everyday language with which to talk about intimacy and wishing that clinicians routinely initiated sexual health counselling.

> It is difficult to express oneself … correctly … […] just talking about it [the condition] with a partner. Because I usually don't talk about it, so it kind of… […]. And I was never prepared for how I could talk about it with others, so it is maybe an area where I am a bit unsecure. (Ellida, XX,female, 16–20 years)

Talking about private and intimate parts of the body appeared to be difficult, especially for the youngest participants. Some participants used words such as "thing" when talking about their genitals, thereby indicating a reluctance to use medical terms and/or a lack of everyday language.

Four participants had not had sexual experience with a partner and two of them had never been in a stable romantic relationship. Moreover, they gave the impression that they were unwilling to talk about sexual intimacy during their interview. In fact, their answers to the question regarding their thoughts about future intimacy were experienced by the interviewer as both guarded and dismissive.

Prospects of fertility

Having a body that may potentially have problems conceiving a child represented a significant experience reported by all the female participants. Three female participants reported being unable to have biological children and stated that they had accepted it. Having other important interests in life and considering alternative ways of being a mother, such as adoption or egg donation, were reported as reasons for the acceptance of infertility. However, the acceptance of infertility was not always as easy as it first appeared, as illustrated by Nora's ambivalent feelings concerning this theme when the interviewer asked her if fertility was something she thought about: "No, not really… Sometimes, when I lie in bed maybe, alone. Maybe I think a little bit about it. I can get a little sweaty … or kind of get… almost like a panic attack. I can get that" (XY,female, 21–26 years). For her part, Aurelia indicated that acceptance might become more difficult as she gets older: "and over the last year, the wish for my own children has increased, but I haven't been sad about it, because I have thought since I was 15 that 'well, it is just how it is" (XY,female, 16–20 years). All the participants for whom this was an issue reported talking to a partner

about possible or certain infertility to be difficult. In addition, the participants described the disclosure of fertility issues to become more relevant and also more difficult as they got older.

One-third of the participants (all females) were unsure about their prospects of fertility. A lack of information or discrepancies in the information provided to them were experienced as a significant strain. Susannah related being initially told that she could not conceive a child, although she was later informed that she had a good chance of fertility. She described a long journey to receiving accurate and adequate information after not being taken seriously regarding fertility issues: "It actually took six years from the time I started requesting information until I was taken seriously! Because I was then old enough to be considered a possible parent" (XX,female, 21–26 years).

None of the male participants reported thinking about their future fertility, nor did any of them problematize the issue, although the oldest male participant did talk about his prospects of having a family when he was older.

Discussion

This study sought to develop an in-depth understanding of the experiences of AYA with DSD by focusing on their experiences of intimacy and sexual health. The key findings of the study revealed that the participants who were sexually active described intimacy and sex as being problematic in terms of both their genital functioning and their genital appearance. Furthermore, having limited knowledge about their condition and lacking everyday language affected the participants' feelings of differentness, which combined with their feelings of stigma. The female participants expressed ambivalent feelings regarding infertility, which influenced their decision to discuss the topic with their partner.

The participants in this study were clearly engaged in an ongoing process of finding acceptance and trying to understand their sexual health needs. Some reported sexual well-being while others reported having no sexual experience with a partner, which confirmed sex and intimacy to be highly individual amongst AYA with DSD. Ongoing functional difficulties (penis/vagina) and atypical genitalia were described as factors contributing to the feeling of not being normal. The related functional difficulties included a reduced sex drive, short and/or narrow vagina, penis dysfunction (e.g., due to curvature), and hormonal imbalances due to medication or DSD symptomatology. These findings are in line with previous studies showing that painful intercourse and dissatisfaction with genital function can have a negative effect on sexual health (Köhler et al., 2012).

Having genitals that appeared more "normal" was a topic raised by all the participants in this study that affected their genital development (most XX,DSD females and XY,DSD males). Genital surgery can help to promote a feeling of normalcy in some individuals with DSD (Boyle et al., 2005). Normalization has previously been identified as a common coping strategy among individuals living with multiple conditions (Sanderson et al., 2011). In fact, one of the six distinct normality typologies proposed by Sanderson et al. (2011) is "struggling for normality, presenting a normal life whatever the cost." In this study, the participants expended a lot of mental resources on feeling different and emphasized how striving for normalization entailed adjustments. Some were even willing to pay for a chance at normalcy. An individual's experience of their deviant appearance and the disruption to their bodily identity are both influenced by cultural norms concerning how a normal and attractive body should appear (Eagly et al., 1991; Toombs, 1995). Given their key role in shaping how individuals feel about themselves, appearance and body satisfaction likely represent the most important contributing factors to self-esteem (Thompson & Kent, 2001). Individuals with a visible difference may experience problems in their social life (Tiggemann, 2001). Moreover, it is challenging to reveal a deviant appearance to others because it can result in negative reactions from others and discrimination. Some of the participants in this study indicated genital appearance to be more important than function.

The AYA who participated in this study described applying the coping strategies of hiding or not revealing their DSD condition to friends and sexual partners. As individuals with DSD have a concealable condition, they might experience being stigmatized when compared with traditional conceptions of "normal" male and female bodies. This might lead to them anticipating stigma if their condition became known, thereby resulting in concealment (Quinn & Chaudoir, 2015). A prior study on DSD determined dissatisfaction with sex life to be associated with traumatic sexual experiences, stigma, and social anxiety (Wisniewski et al., 2019). Other studies on DSD have found that sexual health is associated with the physical condition itself, use of medication, mental health history, body image, gender, age, psychological support, culture, and social media (Hegde et al., 2022; Meyer-Bahlburg et al., 2018; Wisniewski et al., 2019).

Leder (2022) emphasized how dealing with an ill and impaired body requires the application of different strategies during the healing process. He referred to some healing strategies as being intended to "free oneself from the body" (e.g., *ignoring*, *refusing*) and presented others as being designed to "embrace the body" (e.g., *accepting*, *befriending*). These strategies were clearly apparent among the AYA with DSD who participated

in our study. For example, some participants described how their sexual well-being depended on their partner's response and explained how *ignoring* bodily problems and focusing more on strengths (e.g., a positive or neutral reaction from a partner) made sex appear less threatening. In addition, despite the negative feelings associated with being infertile, the female participants seemed to be moving forward in life, which indicated that they were *refusing* "to give in to the body as a controlling factor in their social, emotional or professional lives," as described by Leder (2022, p. 142). In this study, the female participants who were infertile or had problems with menstruation used strategies for acceptance. Nora's description of her tension captured the struggle involved in *accepting* reality: "Maybe I think a little bit about it … almost like a panic attack." Moreover, *befriending* entails an attitude of bodily care best illustrated by Henrik, who initially felt vulnerable in relationships because his penis looked different but decided "It is not that important at all really."

Need for information clarity

The findings of recent studies have suggested that healthcare professionals remain reluctant to talk about sexual issues to AYA with DSD (Callens et al., 2021; Wisniewski et al., 2019). Most participants in this study expressed the need for individualized information and consultation concerning fertility and future parenthood, potential treatment options, and sexual functioning. This study also found that AYA need everyday language with which to communicate about their DSD condition to healthcare professionals. A critical review of the literature regarding the psychological well-being of those with DSD found that if healthcare professionals use only medical terminology, it leads to affected individuals believing that they can only talk to medical professionals (Roen, 2019). Uncertainties about genital surgery and fertility were reported to be burdensome in the present study, especially when accompanied by inconclusive information. Thus, clear communication is particularly important when AYA require healthcare support for the emotional challenging they experience.

Recent studies have focused on fertility issues among AYA with DSD (Corona et al., 2022; Papadakis et al., 2021). For instance, Corona et al. (2022) revealed how support from family, clarity of information, and support from individuals with similar diagnoses were all important factors when dealing with fertility issues. Fertility was an important issue for the female participants in the present study, who reported experiencing inconsistent information and feeling that their concerns were not taken seriously or prioritized. Several participants focused on alternative routes to parenthood such as adoption or surrogacy. However, the male participants in this study did not share experiences of fertility. This might be because fertility was not addressed as an issue during consultations, because they lacked information about potential difficulties, or because they were not willing to share information due to their young age. A prior study reported lower fertility and less satisfaction with sex life in males with severe form of hypospadias when compared with control groups (Örtqvist et al., 2017). Fertility is shown to be dependent on the type and severity of the condition, frequency in partnership, and postoperative complications of genital surgery (Asklund et al., 2010; Skarin Nordenvall et al., 2020; Örtqvist et al., 2017).

Clinical implications

Heterogeneity is a key feature of DSD. Indeed, most participants in this study emphasized the importance of receiving individualized medical information and consultation. Since the publication of the first consensus statement on DSD in 2006 (Hughes et al., 2006), the importance of full disclosure of medical information to individuals with DSD has been emphasized. This change in the approach to medical consultation has presented opportunities to broadly discuss healthcare needs with parents, children, and AYA with DSD (Brennan et al., 2012; Roen, 2019; Wisniewski et al., 2019). Based on the present findings, further understanding is required regarding sexual well-being and different aspects to think about in those with DSD from the clinical perspective. Today, healthcare professionals need to initiate conversations about sexual health and sensitive issues, and they need to become better at letting AYA know that it is fine to talk about sex. Moreover, healthcare professionals need to stress to AYA with DSD that their problems are not rare, irrelevant, or untreatable. Among the most challenging issues for the female participants in this study were their prospects of fertility, early menstruation, or absence of menstruation. The participants also raised concerns about acceptance among healthcare professionals and the provision of information about genital surgical treatments. In general, the participants expressed positive views about potential cosmetic surgery as part of the treatment plan for a "normal" genital appearance. None of them had experienced any "disapproving gaze" or negative remarks from healthcare professionals, although the emotional reactions stemming from having a functionally and/or visibly different body might lead to feeling different or not "normal".

Strengths and limitations

In this study, the researchers attempted to recruit a heterogeneous sample from two multidisciplinary clinics at two university hospitals and in collaboration

with a national competence centre related to DSD situated at one of the hospitals. The sample size is considered adequate for a qualitative phenomenological study. The diversity of diagnoses might limit the transferability of the findings. However, the explorative and qualitative nature of the study gave rich and nuanced data that represented universal life experiences. As such, the findings may have utility in similar situations and across contexts (Shenton, 2004). However, findings should be interpreted with caution due to the heterogeneous nature of participants (Braun & Clarke, 2019). Further, even though an interpretive phenomenological approach gives an insight in the lived experiences of AYA with DSD, other possible phenomena related to sexual health might be revealed in other cohorts. Future studies may look to expand the present findings to other settings and to include experiences of patient advocacy groups or activists. In terms of dependability and trustworthiness, the findings of this study are reinforced by the transparency of the analysis (Shenton, 2004).

A limitation of the study is that the interviews did not cover sexual activity in detail (e.g., frequency, type of activity, partner's gender). This is because the participants guided the direction of the interview. As some AYA may be unwilling to talk about detailed activity unsolicited, researchers could facilitate for those who wanted to elaborate on this in more detail when ethically appropriate, thorough preparation of the participants in the initial information conversations or in the invitation letter.

Conclusion

The findings of this study provide an in-depth understanding of the experiences of AYA with DSD regarding sexual health and well-being. Three major themes emerged in this study, namely the importance of being "normal," communication, and prospects of fertility. The sexual experiences of AYA with DSD are diverse and likely culturally dependent. The process of finding acceptance occurs outside of and beyond medical settings, although the understanding of AYA with regard to sexual well-being remains focused on medical or surgical treatments. Fear of stigmatization and lack of everyday language has the potential to both complicate communication with healthcare professionals and others. Clinicians may consider to begin by enabling AYA with DSD to express themselves with respect to their unique healthcare needs and to voice their own perspective on sexual well-being. Fertility/infertility issues concerned the female participants in this study, who felt that healthcare professionals do not sufficiently acknowledge such concerns. This finding suggests that clinicians need to find a way to talk to AYA of both genders about fertility.

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Notes on contributors

Line Mediå RN, is a PhD student at the Faculty of Medicine, University of Oslo, Norway, as well as a nurse/counsellor at the Centre for Rare Disorders, Oslo University Hospital. With a clinical background in specialized healthcare involving congenital malformations and rare disorders, her research interests include chronic illnesses, transition phases, disclosure, stigma, diversity of sex development, and congenital malformations. Her recent research has focused on the life course of individuals living with DSD, either as adults with DSD or as parents of children with DSD.

Solrun Sigurdardottir is a licensed neuropsychologist working at the Centre for Rare Disorders, Oslo University Hospital, Norway, since 2019. She completed her Ph.D. thesis on traumatic brain injury in 2010 at Sunnaas Rehabilitation Hospital in Norway. She was a post-doctoral researcher in 2012-2015 at the University of Oslo in Norway. She has 25 years of experience specializing in adults with acquired brain injuries. Publications (ca. 60 articles) have focused on neurocognitive outcomes and psychological consequences after traumatic brain injuries e.g., executive functions, olfactory function, depression, anxiety, post-concussion syndrome and posttraumatic stress disorder. More recently, her research work has focused on rare disorders and quality of life. Additionally, she is a supervisor and bi-supervisor for several Ph.D. candidates and has a history of managing specific projects that have resulted in multiple publications.

Lena Fauske is a researcher working at The Norwegian Radium Hospital, Oslo University Hospital (NRH OUH) cancer clinic since 2012 and holds a position at the University of Oslo (UiO), medical faculty as an associate professor. She completed her PhD, "Cancer more than a disease" at the UiO in 2016. At the NRH OUH she researches the patient perspective on cancer, and she teaches qualitative methods at UiO and supervises master's and PhD students. The publications have focused on cancer survivorship in sarcoma patients. Her research also involves patients with other metastatic cancers, fatigue in young cancer survivors and

changes in body image among others. She has participated in the research group Society, health and power (SHEP) since 2012. Since 2019 she is a member of EORTC quality of life sarcoma research group.

Anne Waehre is head of the National Treatment Service for gender incongruent children and young people at Oslo University Hospital since 2017. She previously worked as a pediatrician at Karolinska Hospital. She currently has a post doc scholarship related to gender incongruence in children and young people. She is also a supervisor for several research fellows in the areas of gender incongruence and diversity of sexual development.

Authors' contributions

LM contributed to the conceptualization, recruitment of participants, methodology, interviewing, analysis, writing-original draft, and writing-review/editing.

SS contributed to the conceptualization, methodology, analysis, writing-original draft, writing-review/editing, and writing the application for funding (Foundation DAM).

LF contributed to the conceptualization, methodology, interviewing, analysis, writing-original draft, and writing-review/editing.

AW contributed to the conceptualization, recruitment of participants, methodology, analysis, writing-original draft, writing-review/editing, and writing the application for funding (Foundation DAM).

All authors read and approved the final manuscript. All of the co-authors approved the submission of this work for publication.

Data availability statement

Given the qualitative nature of this study, the generated datasets are not publicly available due to participant confidentiality issues.

ORCID

Line Merete Mediå http://orcid.org/0000-0002-4976-6643
Solrun Sigurdardottir http://orcid.org/0000-0002-7194-3570
Lena Fauske http://orcid.org/0000-0002-8069-1221
Anne Waehre http://orcid.org/0000-0002-5457-5606

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III

Dilemmas Faced by Health-Care Professionals Regarding Treatment and Differences of Sex Development: A Qualitative Study

Abstract

Background: Surgical interventions for individuals with Differences of Sex Development (DSD) remain controversial, necessitating shared decision-making among patients, caregivers, and health-care providers. A lack of evidence in support of, for deferring, or for avoiding surgery complicates the decision-making process. Moreover, there is limited research on health-care professionals' perspectives within this context. This study explores health-care professionals' perspectives on decision-making in DSD-related surgeries and the dilemmas they are facing in this process.

Methods: This qualitative study involved 14 health-care professionals integrated into or collaborating with multidisciplinary DSD teams. They participated in three focus group interviews. Transcripts were reflexively and thematically analyzed

Results: Three overarching dilemmas shed light on the intricate considerations and challenges that health-care professionals' encounter when guiding patients and caregivers through surgical decision-making processes in the context of DSD. The first theme describes how shared decision-making was found to be influenced by fear of stigma and lack of evidence-based practice when navigating the child's and caregivers' needs. The second theme illuminated dilemmas due to a lack of evidence-based practice. The third theme described balancing the interplay between concepts of normality, personal experiences and external expectations. The core concepts within each theme were the dilemmas health-care professionals face during consultations with caregivers and affected individuals

Conclusion: Health-care professionals perceive the shared decision-making process to be demanding. Increased knowledge on adults and the consequences of performing or withholding surgery, alongside with the use of Shared decision-making tools may be beneficial.

Keywords

Differences of sex development; Intersex; Surgery; Shared decision-making; Dilemmas; Qualitative

Abbreviations

Differences of sex development (DSD)

Focus group interviews (FGIs)

Multi disciplinary team (MDT)

Highlights

- DSD-related surgeries are controversial and subject to debate
- Health care professionals grapple with dilemmas during decision-making, as revealed in focus group interviews.
- Shared decision-making was found to be influenced by fear of stigma and lack of evidence-based practice
- Health care professionals face dilemmas in the decision-making process

Introduction

Differences of sex development (DSD) represent a heterogeneous group of congenital conditions that cause the development of the genitals, hormones, or chromosomes to differ from traditional conceptions of male and female bodies (Cools et al., 2018)]. DSD necessitate individuals, their caregivers, and health-care professionals (HCPs) making decisions about medical treatment and, for some, different types of surgeries. The conditions categorized as DSD are rare and vary in terms of the severity, degree of complications, psychological impacts, and treatment needs and possibilities (Hughes et al., 2006). Moreover, DSD may be associated with stigma and controversy (Lampalzer et al., 2020), which may adversely affect individuals' physical and psychosocial health, indicating the need for individualized care and long-term follow-up (Lee et al., 2016). Multidisciplinary teams should be involved in medical treatment, surgical management, and follow-up care for those born with DSD (Lee et al., 2016).

DSD-related surgeries are procedures performed on the external genitalia (e.g., male genital reconstructive surgery, clitoroplasty) or internal reproductive structures (gonads) (Gardner & Sandberg, 2018). DSD-related surgeries, and surgical interventions on female external genitalia in particular, are controversial and subject to debate amongst HCPs and human-rights activists (Lee et al., 2016). In 2006, a consensus statement was published to improve the management of individuals with DSD (Hughes et al., 2006). In the statement, there were proposed suggestions for improving DSD care, which required an experienced multidisciplinary team, open communication and a more cautious approach to early genital surgery intended to alter the sexed appearance. Uncertainty regarding the timing of surgery, lack of evidence in support or rejection of surgery, and the child's right to decide complicate the decision-making process and a debate on the

best approach is ongoing (Bennecke et al., 2021; Flewelling et al., 2022; Lampalzer et al., 2020; Roen, 2019).

Making decisions concerning DSD-related surgeries can prove problematic for individuals with DSD, their caregivers, and HCPs (Bennecke et al., 2021; Flewelling et al., 2022; Hegarty et al., 2021; Kremen et al., 2022)]. In general, shared decision-making is recognized to have three essential elements: acknowledging that a decision is required, knowing and understanding the best available evidence, and incorporating the patient's values and preferences into the decision (Légaré & Witteman, 2013)]. Some studies highlight the importance of shared decision-making in DSD care (Siminoff & Sandberg, 2015)], although few studies explore the issue of communication regarding surgical interventions between HCPs and individuals with DSD and their caregivers. Additionally, research suggests that caregivers do not always recognize their part in the decision-making process due to perceiving surgery as necessary (Alderson et al., 2022; Crissman et al., 2011). The literature describes how HCPs may find the decision-making process challenging, points to difficulties with communication and how parents might have difficulties of grasping the complexities with DSD and treatment options (Roen, 2019; Suorsa-Johnson et al., 2022)

The discussion regarding the indications, timing, procedures, and outcome evaluations for DSD surgery is ongoing (Lee et al., 2016). Research into HCPs' perspectives on surgical practices and possible dilemmas may provide useful insights into the shared decision-making process and, therefore, improve health outcomes for individuals with DSD (Kremen et al., 2022). Hence, the overall aim of the present study was to describe HCPs' perspectives on decision-making regarding DSD-related surgeries, and to explore how dilemmas regarding communication with patients/caregivers, and how the use of evidence influenced in surgical decision-making.

Material and methods

We adopted a qualitative design and used focus group interviews (FGIs) to address the study's aim (Kitzinger, 1995). FGIs are useful when aiming to achieve a deeper understanding of participants' experiences, attitudes, or views, as the dialogue and interaction between participants can provide access to their thoughts and perceptions on topics being studied. Also, as the creation of data takes place within the group it can foster a deeper level of exploration (Kitzinger, 1995). In addition, to ensure patient and public involvement, a reference group of AYA (three females and one male) with personal experiences of living with different DSD was established.

Recruitment and participants

The healthcare of children with DSD under 18 years is organized by two regional multidisciplinary DSD teams (MDTs) in Norway comprised by health-care professionals with a range of specialist backgrounds, aiming to provide holistic, patient-centered, and individualized care. The MDTs set up monthly patient-clinics where they meet individuals with DSD/intersex from 0-18 years and their parents. In addition, children, young people and adults are followed up by pediatricians or endocrinologists at local, or university hospitals. The two MDTs arranges semi-annually national reference network meeting. All HCP (n=9) who attended the meeting autumn 2021 were invited to participate in the study, and accepted the participation in FGIs. Additionally five HCPs were prohibited from participation in the national network meeting due to practical reasons, and an extra interview was arranged after the meeting to accommodate this. A total of fourteen HCPs participated. Eight participants were females and six were males, including medical doctors with specialty in endocrinology,

genetics, pediatrics, adolescent & adult gynecology, child- and adolescent psychiatry, pediatric urology, pediatric surgery, plastic surgery and nursing. Their age ranged from 40 – over 70 years (mean = 52.1 years), while their years working with DSD ranged from 1–30 years (mean = 9.8, median=8). The interview guide was designed to elicit accounts of the participants' experience with the decision-making process on DSD-surgery, challenges related to surgery, their involvement in counselling patients and caregivers about surgery, how to support the shared decision-making process and what knowledge was lacking. The participants were able to elaborate on and defend their views if, or when challenged and to share issues raised by the interviewer and other participants (Johnson, 2014). The FGIs were conducted on a face-to-face basis, and their duration ranged from 77–100 minutes. One HCP participated by phone in one of the FGIs.

Data collection

The data were collected in three FGIs, with four to five participants in each interview with various professions and geographical affiliation. The FGIs were conducted during November–December 2021. Two researchers (LM and AW) led the FGIs, accompanied by a moderator (SS and a colleague). The interviews were conducted in Norwegian, audiotaped, and transcribed verbatim by LM. Reflexive summaries were written by the interviewers and moderators and included in the analysis (Neuzil et al., 2022). Measures were made to ensure that each participant got the chance to talk, and individual opinions were encouraged.

Data analysis

The transcripts were first independently assessed by LM and then collaboratively by the researchers to ensure the use of relevant data and to provide a rich analysis. The data analysis was drawn on Braun and Clarke's six-stage process (Braun & Clarke, 2006)

and the principles of reflexive thematic analysis (Braun & Clarke, 2019). First, the transcripts were coded (and recoded) inductively by hand, based on the participants' own words. The impact of the group setting and the interactions that occurred influenced the co-construction of meaning (Wilkinson, 1998). The group dynamic facilitated openness and disclosure as the participants asked follow-up questions relevant to their practice and disclosed differences in practice between institutions (Wilkinson, 1998). Next, the researchers searched for categories, similarities, and divergences in the data. Finally, the researchers agreed on the themes that represented the findings. The researchers assumed a reflexive attitude toward the participants, the interview transcripts, and when interpreting the results, acknowledging that the interviewers asked follow up questions not listed in the interview guide, which might have influenced the results (Wilkinson, 1998). To illustrate each theme, the researchers provided a selection of quotations, which were slightly revised to improve the readability, in line with Kvale and Brinkmann (2009). Quotes from the interviews were translated from Norwegian into English. In the results, an arbitrary number was assigned to each participant (e.g. P3), and a focus group number (e.g. I2) are reported to increase readability (e.g. I3 – P1). The researchers' relation to the topics and data diverged from close to distant, which strengthened the study's rigor (Clarke & Braun, 2021).

In the presentation of the findings labels *few, some* and *most* refers to how many of the group members raised or discussed a particular theme (Krueger, 1997). Extensiveness is rated as '*few*' if a theme was mentioned by 1-4 group members, as '*some*' if it was mentioned by 5-9 group members, and as '*most*' if it was mentioned by 10 group members or more (Slater & Tiggemann, 2010).

Ethical considerations

This study complied with the principles of the Declaration of Helsinki. It was approved by the Norwegian Regional Committee for Medical Research Ethics, South-Eastern Norway (approval #79444) and by the Data Protection Officer, Oslo University Hospital (approval #7000898). All participants got oral and written information about the study and signed a consent letter. To preserve anonymity each participant got a personal identification number in the transcripts. All data (audio files, field notes, and transcripts), were confidential and securely stored. To ensure rigour of the study the Consolidated Criteria for Reporting Qualitative Studies (COREQ) was followed (Tong et al., 2007), the checklist is available as supplementary material (Appendix A).

Results

The participants described various dilemmas affecting decisions concerning DSD-related surgeries. The dilemmas were categorized into three main themes: a) navigating the child's and caregivers' needs, b) dilemmas due to a lack of evidence-based practice, and c) normality – personal experiences and external expectations.

Navigating the child's and caregivers' needs

The caregivers' expectations regarding the outcomes of surgery and the child's right to make decisions about their body after reaching maturity were considered among the most challenging aspects of surgical decision-making. The participants reported arguing in favor of deferring female genital reconstructive surgery for young children in their initial communications with caregivers. Yet, some HCPs had the impression that caregivers found it difficult to adopt a "wait-and-see" attitude and so opted for surgery to make genitals look like most others', in the hope that it would reduce potential negative experiences e.g. in kindergarten, related to having visible different genitals.

The interviews revealed differences in practice where some described conducting a few female genital reconstructive surgeries, and others elaborated on how they had stopped doing this procedure due to the change in practice. However, there were no difference in practice regarding the removal of the inner gonadal structures and male genital reconstructive procedures.

> "*Regarding the girls, we have stopped operating on the virilized girls. I think the last clitoroplasty we did was exactly 10 years ago. So now they grow up more or less virilized*." (I3-P3)

These differences were exemplified by discussions in the FGIs regarding parental expectations.

> "*I1 - P1: We haven't had parents pushing for [early genital reconstructive] surgery to be done either. I1 - P2: Well… I feel that we have. We have quite a few parents who push for [surgery]. […]. Participant I2 – P1: Yes, my impression is that the parents are a bit pushy, they prefer things to look normal*."

Further, HCPs described a wish endorsed by the parents to reduce bullying and stigma, and enable their daughters to wear tight-fitting clothes at the beach without an enlarged clitoris being visible.

> "*And you have the young girls wearing bikinis, having an enlarged clitoris, and the clitoris becomes erect. Or… yes. And it is visible when they wear a gymnastics suit. […]. It isn't always easy for them [the parents], having good arguments for [surgery]*." (I2 – P1)

The presence of good family support was highlighted as crucial in all the interviews in terms of whether the child managed to live with genitalia that deviated from what was perceived as typical for a male or female body. Psychological support and follow-up, as

well as medical guidance during surgical decision-making, were emphasized as important support for family members. However, a few HCPs sympathized with the idea that one way to reduce caregivers' stress and contribute to coping was to "normalize" the child's genitals or to remove gonads that did not accord with the assigned sex.

> "*We know that parents play a big part in affecting their child… And building trust, or simply destroying self-confidence. […]. It may be that we have to listen carefully to the parents' wishes, because we know that some of them might not be able to support their child with a deformity, even if it is very insignificant*." (I1 – P5)

Some HCPs reflected on whether the way information was conveyed might influence caregivers' decisions. For instance, when the HCPs presented the history of moving to a non-surgical treatment concept, they elaborated on earlier clinical practice whereby surgical normalization was common and more accepted. The HCPs felt that caregivers who were presented with such information tended to request surgical options. Hence, the HCPs recognized that they had considerable authority, meaning they had the potential to influence patients' and caregivers' preferences, which could be perceived as an additional dilemma.

> "*I'm thinking a bit about how we convey information to the parents of girls with... with clitoral hypertrophy or abnormal genitalia […]. So, we are the ones who say that we used to do that [surgery], but now we no longer operate because it is agreed that the child should be allowed to decide for oneself. We are the ones informing about previous practice. So, we kind of suggest a solution, but at the same time, we say that they can't have it."* (I1 – P4)

Dilemmas frequently arose in the decision-making process concerning surgery because the participants argued that patients and caregivers should be the primary decision makers. However, the HCPs questioned whether patients and caregivers actually understood all the necessary information regarding the condition, surgical interventions, and debate regarding surgery. Additionally, the HCPs recognized that patients and caregivers may be reluctant to make the necessary decisions.

> "*I am trying to introduce them to this discussion [about genital surgery]. Slightly simplified… or to invite them to investigate a bit for themselves. Take responsibility for considering this for themselves. And they just say: 'Yeah, I'll do what you say,' and this happens repeatedly.*" (I3 – P5)

Dilemmas due to a lack of evidence-based practice

A major issue for the HCPs was how best to reassure and guide patients and caregivers during the decision-making process when evidence-based practice continues to be guided by contradictory literature and a lack of consensus. Current knowledge is based on small sample sizes and a lack of optimal surveillance possibilities when the removal of gonadal tissues is in question. Additionally, a few participants emphasized that, in their experience, the perception of DSD-related surgery as being undesirable was based on anecdotal narratives from some patients. This was problematic in terms of which voices were listened to when shaping current practice.

> "*It will be interesting to see, in 10 years' time, how those who have not had [genital reconstructive] surgery experienced it. Somebody should follow up on that. Because we are aware that we know too little about it. And they are not the ones who speak the loudest in the activist forums*." (I3 – P5)

Others highlighted how it might be problematic for patients to voice contentedness with surgery when disclosure is problematic due to internalized or expected stigma and when

there is little knowledge and understanding of DSD among colleagues and in society in general. Accordingly, both positive and negative experiences of surgery may be undermined.

> "*It is really important to gain more knowledge, for those of us who work with DSD and for outsiders. Even in the department where I work, people don't know the difference between DSD and transgender. They have no idea […]. And I find it very, very unsettling that people know so little about it.*" (I1 – P2)

Some HCPs identified a lack of financial and human resources, as well as an absence of patient registries, as major hurdles with regard to conducting longitudinal and retrospective research among the DSD patient population. Research is necessary to produce the evidence-based knowledge needed during the decision-making process.

> "*For instance, those gonadectomies, […] we have too little knowledge about what actually happens to the gonads in several of these DSD conditions. We need to do something about this lack of knowledge at some point*." (I2 –P2)

The participants described gonadectomies as interventions to which they sought to strike the correct balance in terms of information dissemination. Some HCPs found it challenging to provide conflicting medical information, for example, discussing the beneficial effects of gonadal hormone production while also explaining the increased cancer risk. All the HCPs recommended giving patients and caregivers time to process the provided information as part of the shared decision-making process.

> "*Then there's this thing with Swyer, we've had several cases of cancer. We need time to provide them [the patients] with this information [about the risks and benefits of gonadectomies], so that they can understand this… For them to*

> *understand the risk and to be part of the decision-making process regarding gonadal removal*." (I3 – P5)

However, some participants recognized that more experienced and competent colleagues could communicate a greater level of confidence in their evaluation of surgery by drawing on a broader clinical picture in the decision-making process.

> "*We don't have a registry, nor data with long-term results. A lot of the information we provide and the follow-up care are built on clinical experience. So, if we are going to do this in a proper way, we need to conduct a type of follow-up procedure that we can extract results from. […]. From a short- and long-term perspective.*" (I3 – P5)

Normality – personal experiences and external expectations

Participants brought forth dilemmas regarding navigating between a universal need for normality and expectations from others then caregivers. These dilemmas became evident when HCP described arguments in favor of DSD-related surgical intervention and arguments for postponing or not performing surgery and when these arguments were conflicting. Notably, the child's best interests seemed to be the top priority, but the best path was not always evident. For instance, genital reconstructive surgery to facilitate boys being able to stand and pee, a normalized appearance and function of genitals to facilitate transitions (starting kindergarten, starting school, puberty, initiating a romantic relationship), and letting the assigned sex better match the child's internal physical features (gonads or chromosomes) were discussed as arguments favoring surgery. By contrast, the arguments for postponing or not performing surgery included the expectations of the international medical, psychological, and activist communities.

> "*Nowadays, we don't do [early female genital reconstructive] surgery anymore, because there is consensus that the child should be given the opportunity to be involved in the decision-making*." (I1 – P3)

Dilemmas arose when the HCPs needed to weigh different values against each other, for example, letting the child attend kindergarten with a "traditional" genital appearance so that the changing of diapers would not be a situation of exposing visible genital differences versus letting the child lead the decision-making when old enough.

Another factor complicated the wish to follow international consensus. This was when HCPs experienced that adolescents and young adults were pleased with surgery being done in early childhood, or when the patients did not problematize the issue with DSD-related surgery.

Other arguments were positive feedback from adults who had undergone early surgery, and an improvement in surgical techniques. These experiences were particularly evident regarding genital reconstructive surgery when an individual was born with proximal hypospadias. Most participants considered proximal hypospadias surgery to be less controversial than female genital reconstructive surgery.

> "*Obviously, it's totally forbidden to operate on girls. However, for under-virilized boys, who one assumes will reach a male identity, early operations are totally accepted. Just as long as you have the parents' consent*." (I3 –P3)

The HCPs experienced that adolescents did not problematize hypospadias surgery despite the complications that might arise afterwards (e.g., fistulas). The participants were under the impression that adolescents were content with the decision made regarding surgery on their behalf when they were infants. However, they discussed how the lack of follow-up after 16 years of age was a possible reason for the lack of

problematization concerning DSD-related hypospadias surgery, as problems may occur later in life.

Discussion

This qualitative focus group study explored the complex dilemmas faced by HCPs who were familiar with DSD, and who were involved in decisions related to Differences of Sex Development (DSD)-related surgeries. We identified several overarching dilemmas concerning the decision-making process. Most notably, shared decision-making was still, almost two decades after the consensus statement, found to be influenced by fear of stigma and lack of evidence-based practice. HCPs described major dilemmas they faced during the decision-making process and in guiding affected individuals and caregivers, for example, how to best support and communicate with caregivers, and how to address uncertainties related to surgeries. Balancing the interplay between concepts of normality, personal experiences and external expectations became evident. These themes shed light on the intricate considerations and challenges that HCPs encounter when guiding patients and caregivers through surgical decision-making processes in the context of DSD.

In the last two decades, health care of children and adolescents with DSD has shifted from a focus on "normalizing" surgery as the solution to the child's difference towards a family-centered health care, where the psychosocial and family-educational needs receive more attention (Suorsa-Johnson et al., 2022). Consistent with past literature, our findings underpin that HCPs still face dilemmas and contrasting interests when counselling individuals born with DSD and their families (Chan et al., 2020). An example of contrasting interests is when the surgical options were explained, the caregivers of infants and young children tended to perceive DSD-related surgery as a

"way out" of having a child that challenges "normality" and a way of avoiding the risks of stigmatization and bullying. Prior studies have described the dialogue concerning genital surgeries between doctors and caregivers of children with DSD, revealing that surgeries were often chosen by caregivers to reduce the uncertainty regarding their child's future (Ellens et al., 2017; Timmermans et al., 2018).

When ambivalent messages are delivered by clinicians, such as surgery being described as both beneficial and potentially risky in the longer-term, research indicates that caregivers selectively follow arguments promoting surgery (Timmermans et al., 2018). Furthermore, when HCPs struggle to reconcile the prevalent belief that surgery is necessary, despite being aware of the potential negative consequences, and the focus on the child's right to decide, arguments for deferral can be perceived as unclear by caregivers (Liao et al., 2019; Timmermans et al., 2018). Therefore, information that is perceived as conflicting can complicate informed consent from caregivers and affected individuals. Moreover, reflection on whether HCPs' existing knowledge and treatment recommendations are based on individual experiences, attitudes, or evidence-based knowledge is necessary. Unfortunately, literature on educational interventions is lacking in DSD, but the use of checklists to avoid conflicting information from various providers has proved useful (Graziano & Fallat, 2016). Such a checklist is a valuable tool to address sensitive issues with patients and their parents. Examples of such tools are provided by e.g., Pediatric Surgeons of Phoenix "DSD diagnoses checklists" (Pediatric Surgeons of Phenix, 2023)].

Some HCPs found it challenging to navigate between the child's and caregivers' needs during the decision-making process, with caregivers being perceived to be anxious about postponing or not pursuing genital reconstructive surgery for their child. The literature supports the notion that surgeries to correct genital ambiguity are, in some

cases, performed to alleviate stigma and distress in both caregivers and affected individuals (Bougnères et al., 2017)]. Furthermore, Kremen et al.'s (2022) recent study of caregivers' decisions to pursue feminizing genital procedures demonstrates that pre-surgical levels of anxiety were lower in the mothers of children who did not undergo clitoroplasty and vaginoplasty than in the mothers of children who underwent such surgery, irrespective of the diagnostic severity. However, a quantitative study of caregivers of children with moderate to severe genital atypia, suggests that the caregivers' anxiety and depressive symptoms decreased over time regardless of their children received or declined surgery, whereas illness uncertainty was predictive of caregiver distress (Roberts et al., 2020). These results indicate that HCPs should focus on affected individuals' and caregivers' reasoning and feelings about decisions regarding surgery, in order to help them explore their own motivations for choosing or declining surgery. Yet, the dilemma of the child's right to decide regarding their body (principle of autonomy) and the dilemma involved in whether the decision maker has fully understood the implications of surgery (principle of informed consent) need to be considered. When the knowledge base is limited regarding the consequences of surgeries, as experienced by our participants, physicians need to question whether the foundations for informed consent are in place.

DSD-related hypospadias surgeries were considered less problematic than female genital reconstructive surgery by most HCPs in this study. The lack of feedback concerning postsurgical regret from patients and giving boys the chance to grow up with a "normal" looking and functioning penis were highlighted as reasons to perform surgery. There might be several reasons for the apparent difference in perspectives regarding male and female genital surgery. First, little surgical regret has been reported in relation to male reconstructive surgery by caregivers (Ellens et al., 2017) or affected

individuals (Flewelling et al., 2022). Furthermore, the gender-wise differential effect of shame and cultural assumptions were suggested as reasons for viewing "normalizing" surgery as an alternative for boys with hypospadias while problematizing female genital reconstructive surgery. Second, the World Health Organization has called for a moratorium on female "normalizing" surgery, although it has not done the same regarding male "normalizing" surgery (Earp et al., 2021). Relatedly, a new Icelandic law states that no permanent changes to sex characteristics, other than hypospadias surgery, can be made while the child is too young to provide consent (Alaattinoglu, 2022), which is in consistency with other countries' laws (e.g. Malta and Germany) (Danon et al., 2023). A moratorium on surgery is also supported by human-rights activists (Human Rights Watch, 2017). While the bioethical literature has discussed the contradistinction between female and male genital surgeries, the medical and psychological literature has focused less on this issue, rendering it less applicable for HCPs (Earp et al., 2021). Roen and colleagues (Roen & Hegarty, 2018) found in a qualitative study that the way information was conveyed was guiding parents' decisions towards or away from the surgery, and that by giving parents a chance to talk with a psychologist, they were empowering parents to say 'no' to hypospadias-surgery. This is one of the few study to question hypospadias surgery.

For the optimization of shared decision-making in clinical perspectives, a well-established knowledge base is required and an understanding of HCPs' dilemmas is warranted. A lack of evidence-based knowledge may heighten HCPs' individual preferences and institutional differences (Légaré & Witteman, 2013). This indicates a need for shared decision-making tools that have been shown to increase patients' knowledge and improve shared decision-making (Siminoff & Sandberg, 2015). Nevertheless, prior studies have identified several obstacles relevant to the shared

decision-making process concerning DSD, including cultural factors (e.g., stigma), when the patient's perspective is important but difficult to obtain due to age, and when the decision makers' choices diverge from the best available medical evidence (Légaré & Witteman, 2013). Even though decision-making dilemmas are still ongoing in DSD-clinics two decades after the first consensus statement was announced (Lee et al., 2006), it became apparent in the FGIs that HCPs need to acknowledge that issues such as stigma, culture differences and the child's age are still relevant when counselling individuals with DSD and their families. A decade after the first consensus statement a report was published to look at changing practices and found modest improvement in some areas, but that genital surgery in infancy remains common (Michala et al., 2014).

This study had several limitations. First, the results only reflect the perspectives of HCPs familiar with DSD. It would have been useful to compare the HCPs' perspectives with affected individuals' views on the same topics, or with the views of caregivers who have participated in the proxy-surgical decision-making process. The study participants represents a small group of HCPs, which might limit the transferability of the findings. However, the explorative and qualitative nature of the study gave rich and nuanced data that represented universal experiences of HCPs. As such, the findings may have utility across contexts (Clarke & Braun, 2021, p. 143). Another limitation is the heterogeneity of the focus groups. Homogeneity within groups is recommended to increase shared experiences (Kitzinger, 1995); however, a diversity in professional backgrounds and places of employment increases the visibility of different perspectives and practices and enhances trustworthiness (Shenton, 2004). Finally, only six of the HCPs conducted surgery, which might have affected some participants' sense of the everyday relevance of the subject under study. Yet, most participants had encountered patients where surgical decisions were or had been an issue. Additionally, the discussion between those

who conducted surgery, and those who did not, did not reveal particularly differences, but rather contributed to the explorative nature of the discussion.

Conclusion

This study highlights the intricate dilemmas faced by healthcare professionals involved in DSD-related surgical decisions. The themes of navigating caregivers' needs, grappling with a lack of evidence-based practice, and balancing external expectations underscore the complex landscape of decision-making in this field. Addressing these challenges requires a multidisciplinary approach that encompasses comprehensive patient education, robust research efforts, and collaborative discussions among medical professionals, patients, and advocacy groups. As the medical community continues to learn and evolve in its understanding of DSD, ethical and patient-centered decision-making should remain at the forefront of clinical practice. This necessitates research on the long-term consequences of undergoing or postponing surgery for those affected. Further, even though decision-making dilemmas are shown in other studies, it is important to acknowledge that the experienced dilemmas HCPs are facing are still relevant in today’s clinical practice.

Increasing knowledge in society on gender diversity in general, and DSD in particular, can help affected individuals and their families to accept their own diversity. However, they also need psychological support, and HCPs who share complete, honest, and unbiased information with patients and families while using shared decision-making -tools.

Data Availability Statement

Given the qualitative nature of this study, the generated datasets are not publicly available due to participant confidentiality issues.

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Conflict of Interest Statement

No potential conflict of interest was reported by the authors.

Ethical approval

This work has been approved by the Norwegian Regional Committee for Medical Research Ethics, South-Eastern Norway (approval #79444) and by the Data Protection Officer, Oslo University Hospital (approval #7000898).

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Appendix 1: Interview guide. Study I

Intervjuguide Unge voksne

INNLEDENDE INFORMASJON

Innledningsvis vil jeg gi informasjon om:

- Studien
- Intervjuet
- Informert samtykke
- Muligheten til å trekke seg
- Bruk av lydopptaker
- Transkripsjon og sletting
- At data er avidentifisert
- I intervjuet kan det dukke opp tema og spørsmål som kan oppleves som private eller sensitive. Det vil vi ta hensyn til i intervjuet.

Deretter vil jeg be om informasjon om:

- Alder
- Jobb/studier
- Sivil status
- Familiesituasjon (bor med foreldre, alene, søsken, egne barn)

BEGYNNELSEN: TIDEN FØR OG DA DIAGNOSE BLE STILT

- Jeg vil gjerne starte fra begynnelsen. Fortell meg om diagnosen
- Hvis sen diagnose: hvordan var det frem mot diagnosen ble stilt? Var det noe som endret seg?
- Kan du huske når du for første gang fikk informasjon om (din kroppsvariasjon)? (Hvem, hvordan, opplevelse?)
- Nå i ettertid, tenker du at du fikk den informasjonen du selv hadde behov for? Eller var det noe informasjon du hadde behov for, men ikke fikk? Vet du hva du kan forvente i fremtiden? Fortell litt om det
- Hva kunne ha blitt gjort annerledes?
- Er det noe du ønsker å dele selv angående (din kroppsvariasjon) før vi går videre?

Sårbare perioder og overganger

Det er lite kunnskap om overganger og sårbare perioder i ditt liv, i deres foreldres liv, og om det å bli voksen med en slik diagnose. Dette vil vi gjerne vite mer om

- Om typiske overganger:
  - Hvordan var det da du begynte på ungdomsskolen? VGS?
  - Hvordan var puberteten for deg?

        - Hvordan var det å dusje med de andre?
        - Å gå fra barnemedisin til voksenmedisin
- Er det noen perioder i livet som har vært spesielle sårbare? Kan du fortelle litt om det?
    - Er det noen rundt deg som kan ha en rolle i slike sårbare perioder? Evt hvordan/hvorfor? Helsepersonell?
- Hvordan har det vært/vil det bli for deg å bli voksen? Hva tenker du er viktig for å leve godt med en DSD?
    - Er det noe du går glipp av pga sykdommen?
    - Noe som oppleves urettferdig?

Tenker du at du er annerledes enn andre på din alder? Ser du på din kropp som annerledes enn andres? Hvis nei, hva med deler av kroppen som vi ikke ser?

Behandling/kirurgi

- Kan du si litt om hvilke oppfølging- eller behandlingstilbud som blir gitt personer med (kroppslige variasjoner)?du har fått?
- Hvordan opplever du behandlingen du har mottatt?
- Noe savnet eller som har vært negativt for deg? Hva har vært bra?
- Kan du fortelle hvordan du har blitt involvert i avgjørelser om behandling? Drøftet valgmuligheter? Samtykke?
- Kan du si noe om hvordan oppfølgingen har vært for deg?
    - I DSD-teamet

I samfunnet i dag diskuteres det en del om hva som er viktig å tenke på når det skal gjøres operasjoner på barn eller unges kjønnsorganer. Vi trenger derfor å lære mer av dere.

- Vil du fortelle litt om hvilken operasjon(er) du har hatt og hvorfor operasjon(ene) er blitt gjort?
- Hvordan det var da for deg?
- Husker du hvilken informasjon du fikk i forkant av operasjonen?
- Kan du fortelle litt om du har blitt involvert i avgjørelser som er tatt om eventuell operasjon, og hvordan du har blitt involvert?
- Husker du om det var noen diskusjoner rundt kirurgi, og hvordan opplevde du evt disse diskusjonene?
- Hvordan tenker du livet hadde vært uten operasjonene, eller om de hadde blitt gjennomført på et senere tidspunkt?
- Vet du om eventuell framtidig operasjon eller annen behandling du skal motta i framtida?

Informasjon

- Hvor opptatt er du av sykdommen din? Hvor finner du informasjon om den? Googler du?
- Hva er viktig for deg i en informasjonssamtale (med en behandler)?

- Kan du fortelle om hva informasjonssamtaler med helsepersonell typisk dreier seg om?
    - Noe du savnet?
    - Endret seg etter at du ble større/voksen?

- (Hvis dette ikke nevnes i første spørsmål:
    - Kan du huske når du første gang fikk informasjon om din tilstanden? Hvordan var det for deg?
    - Kan du huske hvem som gav deg informasjonen, og hvordan det ble gjort? Hvordan var det for deg den dagen/den situasjonen/der og da – da du fikk høre barnets diagnose?
    - Fikk du den informasjonen du selv hadde behov for?

Åpenhet om diagnosen

Nå snakkes det mye om åpenhet og at det er viktig å være åpen, spesielt innenfor psykisk helse.

- Hvordan er det for deg å fortelle om din (kroppslig variasjon) til andre? Hvordan forklarer du (kroppslig variasjon) til andre? Hva forteller du?
- Hvem forteller du det til, og i hvilken sammenheng?
- Er det noe spesielt du forteller om, og noe spesielt du ikke forteller?
- Har…(temaene ovenfor) endret seg over tid for deg?
- Tenker du at andre legger merke til at du er født med denne tilstanden uten at du har fortalt om det?
    - I hvilke sammenhenger opplever du dette?
    - Hvordan oppleves dette for deg?
    - Hvordan håndterer du dette?

Åpenhet innad i familien

Kan du fortelle litt om hvordan dere snakker om diagnosen innad i familien? Er det noe som er vanskelig å snakke om?

- Å balansere informasjon
- Å ta hensyn vs sikre kunnskap om egen kropp/diagnose

Romantiske relasjoner og seksualitet

Nå skal vi stille noen spørsmål om romantiske relasjoner og seksualitet. Mange kan oppleve at det er vanskelig, privat og litt flaut å snakke om egen seksualitet, det er forståelig. Men det pleier å gå bra

Kan du fortelle litt om hvordan det har vært for deg som ungdom og voksen med **forelskelse og romantiske relasjoner**?

    - Har du/har du **hatt kjæreste** eller hatt en romantisk relasjon til noen? (Mann eller kvinne? Jente/gutt?)
    - Forelsker du deg i kvinner, menn, begge deler eller ingen av delene? /legning?

- Har du hatt **seksuell erfaring/erfaring med å være intim** med noen? Vil du fortelle om hvordan dette har vært for deg?
    - Er du seksuelt tiltrukket av kvinner, menn, begge deler eller ingen av delene?
- Opplever du/har du opplevd at din seksualitet er påvirket av (*kroppslig variasjon)*? På hvilken måte?
- Har det oppstått noen **utfordringer** hvor du har måttet søke hjelp? Har du utfordringer i dag som du tenker det kunne vært greit å få hjelp med? Hvordan opplevde/oppleves dette?
- Hva har **bidratt til en positiv utvikling av din seksualitet**?

Til de yngste: Har dere hatt seksualundervisning på skolen? Hvordan var det for deg? Var det noe du tenker som kunne vært gjort annerledes?

Fritid og sosiale erfaringer

- Hva gjør du på fritiden?
- Hva er viktig for deg når du tenker på fritiden din?
- Har diagnosen noen gang påvirket deg til å oppsøke eller unngå enkelte miljøer eller aktiviteter?
    - Hvordan var dette i barndommen? (sykehustid for eksempel)

Psykisk helse

Nå skal jeg spørre deg noen spørsmål om hvordan du føler deg. Med hvordan du føler deg mener jeg følelser som f eks trist, glad eller engstelig og hvordan du opplever å mestre dette.

- I hverdagen, kan det jo gå litt opp og ned med humør og hvordan man har det inni seg.
- Kan du fortelle litt om hvordan du har det i dag?
- Hva er viktig for at du skal ha det bra?

Avslutning:

- Er det noe vi ikke har vært innom, som jeg ikke har spurt om som du tenker det er viktig å dele?
- Hvis du hadde mulighet, hva ville du si til noen som er/var i en lignende situasjon som deg?
- Hvordan har denne samtalen vært for deg?
- Var det dette du trodde intervjuet skulle handle om? Noe spes du hadde forberedt deg på? Gruet eller gledet deg til?
- Hva tenker du har vært avgjørende for at det gikk så bra/ble så vanskelig?

Prober:

- Har det alltid vært slik?

- Kan du si noe mer om hva som var greit, eller vanskelig?
- Har du noen eksempler?
- Er det noe det kunne vært mer av?
- Er det noe det kunne vært mindre av?
- Du sa noe om at du følte/tenkte at det var XXX. Kan du huske at du også hadde andre følelser eller tanker? Har du opplevd at følelser du har hatt har vært motstridende?
- Dette er kanskje litt vanskelig å sette ord på, men jeg vil gjerne/kan du si litt mer om hva du mener med…/hva du tenker på når du sier.../hva xxx betyr

Appendix 2: Interview guide. Study II

Intervjuguide

Variasjon i kroppslig kjønnsutvikling (Bufdir, 2018)

Generell innledning:

Kort informasjon om seg selv.

Gjenta hovedpunktene fra infoskrivet: Litt om prosjektet/studien, informasjon om lydopptak, transkripsjon og sletting, informert samtykke, frivillig å delta, mulighet til å trekke seg og at dataene er avidentifiserte.

Studien fokuserer på mennesker med variasjoner relatert til kjønnskromosomer, kjønnshormoner og/eller kjønnskarakteristikker. Deltakergruppen i denne studien er veldig bred og har forskjellige tilstander og opplevelser. Forskjellige spørsmål er relevant for forskjellige personer, så vi ønsker å undersøke hva som er relevant for deg og ditt liv. Fokuset på dette intervjuet er din egen opplevelse, så ingen svar er rette eller gale.

Har de levert spørreskjema?

Intervjuguide voksne og ungdom (over 16 år)

➢ Personalia

- Alder, bosted, kjønn, sivilstatus, etc.

- **Kan du si litt om hva motivasjonen din er for å delta?**

➢ Informasjon

(Dette er jo en studie om variasjoner i kroppslig kjønnsutvikling. Vi har alle forskjellige måter å snakke om kroppslige variasjoner på, eller vi kan også velge å ikke snakke om det. Når vi må snakke om det så er det forskjellige ord vi kan bruke: noen er medisinske eller diagnosetermer. Noen velger andre ord eller begreper.)

- Hva kaller du *din variasjon/diagnose/tilstand*?
  - *(Er det OK at jeg også bruker dette begrepet, eller er det en annen måte du vil jeg skal snakke om det på?)*
- Hva vet du om (*din kroppsvariasjon)*?
- Kan du huske når du for første gang fikk informasjon om (*din kroppsvariasjon)*?
  - *(Hvem, hvordan, opplevelse?)*
- Nå i ettertid, tenker du at du fikk den informasjonen du selv hadde behov for? Eller var det noe informasjon du hadde behov for, men ikke fikk?
- Hva kunne ha blitt gjort annerledes?
- Er det noe du ønsker å dele selv angående (*din kroppsvariasjon)* før vi går videre?

➢ Behandling og oppfølging

- Kan du si litt om hvilke oppfølging- eller behandlingstilbud som blir gitt personer med (*kroppslige variasjoner)*?

Har du mottatt behandling og oppfølging:

- Hvordan opplever du behandlingen du har mottatt?
    - Noe **savnet** eller som har vært **negativt** for deg? Hva har vært **positivt**?
- Kan du fortelle hvordan du har blitt **involvert i avgjørelser** om behandling? Drøftet valgmuligheter? Samtykke?
- Hvordan har tilbud og oppfølging endret seg **etter du fylte 18 år**? (For eksempel: Blitt fulgt i DSD-team?)
    - Hvordan har du opplevd dette?
- Blir du fulgt opp av noen andre? Hvem er dette eventuelt?
- Noen andre aspekter ved behandlingen du føler kunne vært **ivaretatt på en annen måte** eller **har vært spesielt fornøyd med**, som du vil trekke frem, som vi ikke har vært innom?

For noen består behandlingen av en eller flere typer **kirurgiske inngrep**, gjelder dette deg?

Hvis ja:

- Kan du si litt om **hvilke typer** operasjoner som er knyttet til (*din variasjon)*?
- Kan du fortelle litt om **når** du ble operert og **hvorfor**? Flere operasjoner?
- Hvilken **informasjon** fikk du/dine foresatte på forhånd? Hvilke **valgmuligheter** fikk du/dere?
- I hvilken grad **deltok du i avgjørelsen**? Ble muligheter drøftet med deg? Samtykke?
- Hvordan oppleves **resultatene** av operasjonen(e) du har vært igjennom?
- Hvordan tenker du livet hadde vært **uten operasjonene**, eller om de hadde blitt gjennomført på et **senere tidspunkt**?
- Hvor og hvordan har du blitt **fulgt opp etterpå**?
- Vet du om eventuell framtidig operasjon eller annen behandling du skal motta i framtida?

➢ Åpenhet

- Hvilke **erfaringer** har du med å være åpen om (*kroppslig variasjon)*?
    - **Hvorfor/hvorfor ikke**? Tanker/følelser knyttet til åpenhet?
- Hvordan er det for deg å **fortelle om** din (*kroppslig variasjon)* til andre? **Hvem** forteller du det til, og i **hvilken sammenheng**?
- **Hvordan forklarer du** (*kroppslig variasjon)* til andre? Hva forteller du?
- Har du noen gang opplevd å måtte være åpen om din tilstand **uten eget ønske**?
    - Hvordan skjedde dette (for eksempel synlighet, direkte spørsmål, andre personer har delt det uten ditt samtykke osv.)?
- Tenker du at andre **legger merke til** at du er født med denne tilstanden uten at du har fortalt om det?
    - I hvilke sammenhenger opplever du dette?
    - Hvordan oppleves dette for deg?

- Hvordan håndterer du dette?

➢ Utdanning og arbeid

Våre valg av utdanning or arbeid kan styres av flere ting: Hva vi liker, hvem vi er og hvilke muligheter vi har fått. Vi vil gjerne høre litt mer om du tenker det kan ha vært slik for deg, eller ikke.

- Går du på skole/jobber du?
- Hvordan **opplever du** din skole/arbeidshverdag?
- På hvilken måte blir du **ivaretatt** av skole/arbeidsmiljøet?
- Opplever du **utfordringer** knyttet til (*din tilstand)* i hverdagen?
- Opplever/opplevd å bli **behandlet annerledes** enn andre på noen måte?
- Hva kunne du ønsket at hadde vært **annerledes** i arbeidsmiljøet/skolemiljøet?
- Hvilke **aktiviteter** på skole/jobb synes du det er utfordrende å delta på, og hvorfor?

➢ Fritid og sosiale erfaringer

Valg av aktiviteter og sosiale arenaer kan styres av hvem vi er, hva vi liker, muligheter vi har fått eller andre ting. Noen av oss velger også bort aktiviteter av ulike årsaker.

- Hvordan er du **sosial**? Venner/fritid/organisasjoner/andre **sosiale arenaer**?
- Har faktorer knyttet til (*kroppslig variasjon)* noen gang påvirket deg til å **oppsøke** eller **unngå** enkelte miljøer eller aktiviteter?
    - Hvordan var dette i barndommen? (sykehustid for eksempel)
- I hvilken grad opplever du at (*kroppslig variasjon)* har påvirket relasjoner til andre?
- I hvilken grad opplever du (*kroppslig variasjon)* har preget forholdet til din familie?
- Opplevd å bli behandlet annerledes enn andre (erting/mobbing/diskriminering) på grunn av (*kroppslig variasjon)*? (Hvis ja, beskriv)
    - Hvordan opplever du at dette har påvirket din **identitet og ditt selvbilde**?

➢ Selvbilde

- Hva syns du om deg selv? Hva er du **fornøyd/ikke fornøyd** med? Hvorfor/hvorfor ikke? (for eksempel utseende, egenskaper og kvaliteter)
- Hva tenker du kan ha **formet og påvirket ditt selvbilde** (hvordan du ser og tenker om deg selv)?
    - Hvordan har **behandling/oppfølging** påvirket ditt selvbilde?
    - Hvordan har **sosiale reaksjoner** eller erfaringer påvirket dette?
    - Andre erfaringer som har påvirket?
- Opplever du at selvbildet ditt har **endret seg over tid** (fra du var yngre)?

➢ Psykisk helse

Nå skal jeg spørre deg noen spørsmål om hvordan du føler deg. Med hvordan du føler deg mener jeg følelser som f eks trist, glad eller engstelig og hvordan du opplever å mestre dette.

- Kan du fortelle litt om **hvordan du har det**?
- Hva opplever du er **viktig for at du skal ha det bra**?
- Opplever du/har du opplevd **perioder i livet** er/har vært spesielt **sårbare og vanskelige**, og der oppfølging er mer nødvendig?
    - Hvordan **håndterer du perioder** som oppleves som spesielt vanskelige?
    - Hvordan **håndterer du eventuelle vanskelige følelser**? (Rus eller selvskading noen gang?)
- Hvordan tenker du (*kroppslig variasjon)* kan påvirke/har påvirket hvordan du har det? Hvordan har dette vært for deg?
- Hvordan **påvirker samfunnet** deg angående hvordan du har det? (reaksjoner, medisinsk behandling, sykeliggjøring, diagnostisering, diskriminering o.l.)
- **Hvem snakker du med** når ting er vanskelig?
    - Har du tidligere søkt hjelp hos profesjonelle, eller organisasjoner, personer med lignende erfaringer når du har hatt det vanskelig?

(Ivaretas hvis det kommer frem noe vanskelig i svarene her).

➢ Kjønn og kjønnsidentitet

Dette er en bred studie for mange med forskjellige tilstander og diagnoser. Det finnes derfor også veldig mange forskjellige navn, ord og begreper for å beskrive diagnoser og tilstandene.

- Begrepsbruk: Har du hørt om begrepet **«interkjønn» eller «intersex»**? Hvordan forstår du begrepet «interkjønn»? Hva er dine tanker og følelser knyttet til dette begrepet?
- Mange vi snakker med har/har hatt **tanker eller opplevelser om å føle seg mindre mann eller kvinne** på grunn av sin diagnose eller kroppslige tilstand, eller å ha tanker om sin kjønnsidentitet. Har du dette?
    - Føler du deg som mann eller kvinne, begge deler eller ingen av delene? Hvilket kjønn lever du som?
- Hvordan blir du **møtt av andre** angående kjønn? (Feks: opplevd at andre har stilt spørsmålstegn ved ditt kjønn? Eller sagt at du ikke er fullverdig det kjønnet du opplever å være?)
    - I hvilke situasjoner? Blitt behandlet kjipt/diskriminert på grunn av dette? Fortell mer.
        - *(Kan du noe om hvilke rettigheter du har på grunn av (kroppslig variasjon)? Beskyttelse i diskrimineringslov, mulighet til å endre juridisk kjønn, og til foreldre å kunne endre juridisk kjønn før fylte 6 år).*
- Hvilke tanker har du om hva som kan ha **virket inn på din kjønnsidentitet**?
    - For eksempel: Opplever du at dette har endret seg som følge av behandling, over tid, eller ut fra sosiale reaksjoner og erfaringer?

➢ Romantiske relasjoner og seksualitet

Nå skal vi stille noen spørsmål om romantiske relasjoner og seksualitet. Mange kan oppleve at det er vanskelig, privat og litt flaut å snakke om egen seksualitet, det er forståelig. Dersom du ikke ønsker å svare på alle spørsmålene er det greit.

- Kan du fortelle litt om hvordan det har vært for deg som ungdom og voksen med **forelskelse og romantiske relasjoner**?
  - Har du/har du **hatt kjæreste** eller hatt en romantisk relasjon til noen? (Mann eller kvinne? Jente/gutt?)
  - Forelsker du deg i kvinner, menn, begge deler eller ingen av delene? /legning?
- Har du hatt **seksuell erfaring/erfaring med å være intim** med noen? Vil du fortelle om hvordan dette har vært for deg?
  - Er du seksuelt tiltrukket av kvinner, menn, begge deler eller ingen av delene?
- Opplever du/har du opplevd at din seksualitet er påvirket av (*kroppslig variasjon)*? På hvilken måte?
- Har det oppstått noen **utfordringer** hvor du har måttet søke hjelp? Har du utfordringer i dag som du tenker det kunne vært greit å få hjelp med? Hvordan opplevde/oppleves dette?
  - *(Ved CAIS eller andre diagnoser som innebærer endring i testosteron: «noen kan oppleve en endring i libido/seksuell lyst etter at testikkelvev er fjernet/hormonproduksjonen er endret. Har du merket noen endring?»)*
- Hva har **bidratt til en positiv utvikling av din seksualitet**?

➢ Støtte

- Hvilke **andre aktører** utenfor det medisinske apparatet tenker du er viktig for at din hverdag skal være best mulig? (Feks: NAV, skole, helsesøster, brukerforening, sosiale foreninger, fritidsaktiviteter.. etc.
- Hva slags ideer til **endringer i samfunnet/Norge** har du som muligens kunne gjort hverdagen lettere?

➢ Internett og sosiale medier

Mange mennesker bruker sosiale medier aktivt i sin hverdag. Reaksjoner fra andre på sosiale medier kan bidra til at man får bekreftelse på egne valg, ståsted osv., men noen erfarer også at sosiale medier kan bidra til negative erfaringer og reaksjoner fra andre.

- Hvilke **opplevelser har du med sosiale medier og internett** vedrørende (*kroppslig variasjon)*? (for eksempel finne informasjon, ha kontakt med andre, dele informasjon). Positivt/negativt?

➢ Avslutning:

- Er det noe vi ikke har vært innom, som jeg ikke har spurt om som du tenker det er viktig å dele?
- Hvordan har denne samtalen vært for deg?

Appendix 3: Interview guide. Study III

Intervjuguide for fagpersoner

Variasjon i kroppslig kjønnsutvikling

Fokusgruppe Intervjuguide fagpersoner

- **Generell**
  - Kort introduksjonsrunde for alle deltagerne med:
    - Yrke? Hvor lenge har du arbeidet med denne gruppen? Spesifikke diagnoser/tilstander?
    - Hva omhandler ditt arbeid med denne gruppen?
    - Hvordan opplever du arbeidet med denne gruppen?
    - Hva mener du personlig er det viktigste i arbeidet med denne gruppen?

- **Oppfølgingsbehov**
  - Hvordan tenker dere at oppfølgingen er for denne gruppen? (Hva fungerer? Hva kunne vært annerledes?)
  - Hva tenker dere er pasientenes og/eller de foresattes hovedutfordringer med å leve med sin diagnose? Er det en periode/fase i livet hvor oppfølging er ekstra viktig?
  - Hva er deres inntrykk av pasientenes og foresattes, opplevelser med den samlede oppfølgingen og behandlingen de får?
    - I hvilken grad har du inntrykk av at pasienter og foresatte er åpen om deres opplevelser med behandlingstjenesten?

- **Delt beslutningstaking/involvering i avgjørelser om behandling**
  - I hvilken grad opplever dere at dere har samme agenda som pasienten/foresatte?
  - I hvilken grad tenker dere at pasienter og/eller foresatte involveres i beslutninger om behandling?
    - Er det noen pasienter som ønsker mer involvering fra behandlere i avgjørelser enn andre? Hvilke og hvorfor tror dere dette skjer?
  - Det er en del diskusjoner rundt kirurgi på barn med en variasjon i kjønnsutvikling. Begrep som dras inn i diskusjonen er blant annet: autonomi, «medisinsk nødvendig», samtykke. Hvordan håndteres denne diskusjonen i det norske fagmiljøet.
    - Er det noen som har gjort seg noen erfaringer?

- **Informasjon**
    - I hvilken grad opplever dere at det gis tilstrekkelig informasjon i forkant av behandling (eller annen nødvendig informasjon du må gi)?
    - Hva påvirker hvor mye og hvor god informasjon pasientene får? (Tidspress, forespørsel, annet?)
    - I hvilken grad opplever dere at den informasjonen som gis, blir forstått og mottatt av pasient og foresatte? (Hva tenker dere dette kan handle om?)
    - I hvilken grad må informasjon tilpasses basert på karakteristika ved pasienten (som personlighet, evner, sosial klasse, utdanningsnivå, alder med mer). Hvor lett eller vanskelig synes dere evnt dette er?

- **Kommunikasjon**
    - Hva oppleves som mest krevende i kommunikasjonen med pasienter?
    - Hvordan er kommunikasjonen direkte med barnet/pasienten i DSD-teamet? Hvordan opplever dere foreldrene lar barnet komme til? Hvordan håndteres barnets stemme i klinisk hverdag når barnet har en DSD-diagnose? Hva virker inn på dette?
    - Hva er viktig når man skal snakke med barn og unge med variasjoner i kroppslig kjønnsutvikling? (temaer, fokus, begreper)
        - Hvilket tema er det mest utfordrende?
        - I hvilken setting er det lettest å få til en god dialog?
    - Hvilket fokus/informasjon opplever dere at pasienter og foresatte er mest opptatt av selv?
    - Kan dere beskrive en «vanskelig» pasientsak? Hvorfor?
        - Opplever dere at dere har de verktøyene som trengs for å håndtere såkalte ”vanskelige” pasientsaker? Hva er dette?
    - Forsøk å se for dere en eller to saker du står i eller har stått i, der du opplever å ha hatt en god relasjon til en pasient, og hvor du også har fått gode tilbakemeldinger.
        - Hva er det som har ført til at dette har blitt opplevd som bra?
- **Åpenhet**
    - I hvilken grad opplever dere at barn/ungdom/foresatte har kunnskap om sin/barnets tilstand?
    - Kan dere fortelle litt om hvordan åpenhet om tilstanden blir tatt opp i konsultasjonene?
    - Hvilke råd etterspør foreldrene om åpenhet, og hvilke råd gis?
    - Er det enighet i behandlermiljøet om hvilke råd som gis om åpenhet?

- **Avslutning**:
  - Er det noe vi ikke har vært innom, som jeg ikke har spurt om som dere tenker det er viktig å dele?
  - Hvordan var å være med på dette fokusgruppe intervjuet?

Appendix 4: Information sheet and consent form adults

Oslo universitetssykehus

INVITASJON TIL DELTAKELSE I FORSKNINGSSTUDIE

Vil du bidra til å forbedre livssituasjonen for personer med variasjon i kroppslig kjønnsutvikling? Delta i vår forskningsstudie!

BAKGRUNN OG HENSIKT

Du er registrert med en tilstand som innebærer en variasjon i kroppslig kjønnsutvikling, derfor får du denne forespørselen fra oss.

Vi er interessert i dine erfaringer og **ønsker å lære mer om**:

- Hvordan du har det i hverdagen
- Hvordan du opplever å bli ivaretatt av miljøet rundt deg, på skolen eller jobb.
- Sosialt/venner/familie
- Hva dine erfaringer er med helsevesenet.
- Hva du har fått av informasjon.

Hvorfor vil vi vite dette?
Vi håper at det vi lærer gjennom denne studien kan være med å øke kunnskapen om og forbedre livssituasjonen til personer med variasjon i kroppslig kjønnsutvikling.

Hva er variasjon i kroppslig kjønnsutvikling?
Dette er en samlebetegnelse som viser til de av oss som er født med variasjoner i utviklingen av kjønnshormoner, kjønnskromosomer, kjønnskjertler og/eller kjønnsorgan. Vi vet at gruppene vi ønsker å lære mer om er veldig forskjellige. Mange vil kanskje ikke kjenne seg igjen i begrepene eller beskrivelsene i denne teksten. Dette vil vi ta hensyn til i intervjusituasjonen.

Hvem er vi?
Vi er fagpersoner på Senter for sjeldne diagnoser (SSD) som kjenner tilstandene godt. Vi har fått i oppdrag å gjøre denne studien fra Barne-, ungdoms- og familiedirektoratet (Bufdir).

Når kan jeg delta?
Vi er klare for å treffe deg i perioden mellom mai - august 2018.

Hva skjer med informasjonen jeg gir?
All informasjon du gir behandles anonymt. For å kunne øke kunnskap om dine og andres opplevelser, og hjelpe med å bedre oppfølgingen, vil vi formidle dette i en rapport som lanseres av Bufdir. Vi vil også bruke resultater i artikler til internasjonale tidsskrifter, og som en del av en masteroppgave i Interdisiplinær helseforskning ved Universitetet i Oslo.

HVA INNEBÆRER DELTAKELSE?

Hva må jeg gjøre hvis jeg deltar?

1. **Fylle ut noen spørreskjemaer om hvordan du har det.** Disse spørreskjemaene blir sendt i posten. Du kan velge om du vil fylle de ut hjemme eller om du vil fylle de ut hos oss. Det vil ta cirka 15 minutter.
2. **Intervjusamtale** enten hos oss (på Senter for sjeldne diagnoser ved Oslo universitetssykehus, Rikshospitalet), eller over telefon hvis dette er lettere for deg. Dette vil ta 1-2 timer.

Hvilken informasjon om meg skal brukes i studien?
Hvis du ønsker å delta så kommer vi også til å registrere informasjon om deg fra din journal fra tidligere undersøkelser på sykehuset. Dette kan være informasjon om diagnose og eventuell behandling.

Hva må jeg gjøre hvis jeg er interessert i å være med?
Først må du fylle ut et samtykkeskjema nederst i dette dokumentet og levere det til oss. Etterpå blir du ringt av en prosjektmedarbeider på telefon for å avtale tidspunkt for intervjuet.

MULIGE FORDELER OG ULEMPER

Hva kan være positivt med å delta?
Noen synes det er fint å kunne snakke om sin historie og egne opplevelser og utfordringer med noen som kjenner til tilstanden/diagnosen. Noen synes det er meningsfullt å kunne bidra med sine opplevelser og kunnskap, for på sikt å kunne forbedre sin egen og andres oppfølging og livssituasjon.

Hva kan være negativt med å delta?
Du velger selv hva du vil dele, men noen kan synes at enkelte temaer er vanskelige å snakke om. Hvis dette skjer, så vil vi på best mulig måte ivareta deg. Intervjuet tar 1-2 timer, og tiden brukt kan oppleves som en ulempe.

FRIVILLIG DELTAKELSE OG MULIGHET FOR Å TREKKE SITT SAMTYKKE

Det er selvfølgelig helt frivillig å være med i studien. Hvis du har lyst til dette så undertegner du samtykkeerklæringen på siste side og sender den til oss.

Du kan når som helst og uten å oppgi noen grunn trekke samtykket. Dette vil ikke få konsekvenser for din eventuelle videre oppfølging. Hvis du velger å trekke deg fra studien, kan du kreve å få slettet innsamlede opplysninger, med mindre opplysningene allerede er inngått i analyser eller brukt i vitenskapelige publikasjoner.

HVA SKJER MED INFORMASJONEN OM MEG?

All informasjon om deg som vi får fra spørreskjemaer, samtale og journal, blir lagret avidentifisert på en sikret forskningsserver på Oslo universitetssykehus, med en kodeliste lagret separat og innelåst. Det betyr at informasjonen du gir ikke kan knyttes til deg uten at man har denne kodelisten, som kun leder for studien har tilgang til. All informasjon blir altså lagret sikkert.

Vi har taushetsplikt og alle svarene blir behandlet konfidensielt. Det betyr at ingen andre får vite hva akkurat du svarer på spørsmålene vi stiller. Du kan når som helst få innsyn i hvilke opplysninger som er registrert om deg. Du har også rett til å få endret på eventuelle feil i de opplysningene vi har registrert.

Kristin Feragen (tlf: 23 07 53 57), har ansvar for at opplysninger om deg blir behandlet på en sikker måte. Informasjon om deg vil bli anonymisert eller slettet senest fem år etter prosjektslutt.

GODKJENT STUDIE

Studien er godkjent av Regional komite for medisinsk og helsefaglig forskningsetikk, 2017/2554.

KONTAKTINFORMASJON

Hvis du har spørsmål kan du kontakte:

Charlotte Heggeli
Telefon: 23 07 53 37 / 23 07 53 40
E-post: chhegg@ous-hf.no

Senter for sjeldne diagnoser
Oslo Universitetssykehus HF
Rikshospitalet
Postboks 4950 Nydalen
0424 Oslo

SAMTYKKE TIL DELTAKELSE I STUDIEN

Dersom du ønsker å delta i studien signerer du under og sender denne siden til oss i vedlagt frankert konvolutt.

JEG ER VILLIG TIL Å DELTA I STUDIEN

- ☐ Jeg ønsker å delta i studien
- ☐ Jeg ønsker ikke intervju, men kan godt fylle ut spørreskjema, og få dette tilsendt i posten.

Har du krysset av over, vennligst fyll inn feltene under. Ønsker du ikke å delta, vennligst se bakside.

Sted og dato	Deltakers signatur

Deltakers navn med store bokstaver

Kontaktinformasjon:

Telefon: .....

Adresse: .....

.....

JEG ER IKKE VILLIG TIL Å DELTA I STUDIEN

☐ Jeg ønsker ikke å delta i studien

Det hadde vært nyttig for oss å vite hva som er grunnen til at du ikke ønsker å delta. Denne besvarelsen er anonym og frivillig.

Jeg ønsker ikke å delta på grunn av:

.....

.....

.....

.....

.....

Appendix 5: Information Sheet and Consent form Adolescents and young adults

INVITASJON TIL DELTAKELSE I FORSKNINGSSTUDIE

Å leve med en variasjon i kroppslig kjønnsutvikling

BAKGRUNN OG HENSIKT

Dette er et spørsmål til deg om å delta i et forskningsprosjekt som har til hensikt å utforske de ulike erfaringene som det å ha en variasjon i kroppslig kjønnsutvikling kan innebære. Du er/har vært pasient ved Oslo universitetssjukehus (OUS) eller Haukeland universitetssjukehus (HUS), derfor får du denne invitasjonen. Noen er/har vært fulgt i «DSD-team» på OUS og HUS, som er en Flerregional behandlingstjeneste for usikker somatisk kjønnsutvikling. Prosjektet er et samarbeid mellom Oslo universitetssykehus, Haukeland universitetssjukehus og Universitetet i Oslo (UIO).

Hva er variasjon i kroppslig kjønnsutvikling?
Dette er en samlebetegnelse som viser til de av oss som er født med variasjoner i utviklingen av kjønnshormoner, kjønnskromosomer, kjønnskjertler og/eller kjønnsorgan. Vi vet at gruppene vi ønsker å lære mer om er veldig forskjellige. Mange vil kanskje ikke kjenne seg igjen i begrepene eller beskrivelsene i denne teksten. Dette vil vi ta hensyn til i intervjusituasjonen.

Vi er interessert i dine erfaringer og **ønsker å lære mer om**:

- Hvordan du har det i hverdagen
- Hvordan du opplever å bli ivaretatt av miljøet rundt deg, på skolen eller jobb.
- Hva dine erfaringer er med helsevesenet og den behandlingen som du får/har fått.
- Hva du har fått av informasjon.

Dersom du samtykker til det, vil dine foreldre også inviteres til å delta i studien og fortelle om egne erfaringer. Du har rett til å få innsyn i det som fortelles om deg i disse intervjuene dersom du ønsker det. Da må du ta kontakt med prosjektleder Anne Wæhre (uxwhra@ous-hf.no).

Hvorfor vil vi vite dette?
Vi håper at det vi lærer gjennom denne studien kan være med å øke kunnskapen om, og forbedre livssituasjonen til personer med variasjon i kroppslig kjønnsutvikling.

Hvem er vi?
Prosjektet er en del av en PhD-studie som utføres i samarbeid med ansatte på Senter for sjeldne diagnoser og DSD-teamene på OUS og HUS. Prosjektgruppa består av personer som har erfaring med de ulike tilstandene.

Når kan jeg delta?

Vi er klare for å treffe deg i perioden mellom august 2020 – august 2021.

HVA INNEBÆRER DELTAGELSE?

Hva må jeg gjøre hvis jeg er interessert i å være med?
Først må du fylle ut et samtykkeskjema nederst i dette dokumentet og levere det til oss. Etterpå blir du ringt av en prosjektmedarbeider på telefon for å avtale tidspunkt for intervjuet.

Hva må jeg gjøre hvis jeg deltar?
Du inviteres til en **intervjusamtale** enten hos oss (på Senter for sjeldne diagnoser ved Oslo universitetssykehus, Rikshospitalet), eller hvor det måtte passe deg gjerne i nærheten av der du bor. Det er også en mulighet å gjøre dette per telefon for de som ønsker det. Intervjuet vil ta mellom 1-2 timer.

MULIGE FORDELER OG ULEMPER

Hva kan være positivt med å delta?
Noen synes det er fint å kunne fortelle sin historie, og snakke om egne opplevelser og utfordringer med noen som kjenner til tilstanden/diagnosen. Noen synes det er meningsfullt å bidra med sine opplevelser og kunnskap, for på sikt å kunne forbedre sin egen og andres oppfølging og livssituasjon. Vi håper denne studien vil forbedre helsevesenets tilbud i fremtiden, ved å bidra til at helsepersonell og det aktuelle vitenskapelige miljøet skaffer tilveie verdifull informasjon om hvordan det er å leve med en diagnose som innebærer en variasjon i kroppslig kjønnsutvikling.

Hva kan være negativt med å delta?

Du velger selv hva du vil dele, men noen kan synes at enkelte temaer er vanskelige å snakke om. Hvis dette skjer, så vil vi på best mulig måte ivareta deg.

FRIVILLIG DELTAKELSE OG MULIGHET FOR Å TREKKE SITT SAMTYKKE

Det er frivillig å delta i prosjektet. Dersom du ønsker å delta, undertegner du samtykkeerklæringen på siste side og sender den til oss. Du kan når som helst og uten å oppgi noen grunn trekke samtykket. Dette vil ikke få konsekvenser for din eventuelle videre behandling eller oppfølging. Hvis du velger å trekke deg fra studien, kan du kreve å få slettet innsamlede opplysninger, med mindre opplysningene allerede er inngått i analyser eller brukt i vitenskapelige publikasjoner.

HVA SKJER MED OPPLYSNINGENE OM DEG?

Opplysningene som registreres om deg skal kun brukes slik som beskrevet i hensikten med prosjektet. All informasjon om deg som vi får fra samtalen blir lagret avidentifisert på en sikret forskningsserver på Oslo universitetssykehus, med en kodeliste lagret separat og innelåst. Det betyr at informasjonen du gir ikke kan knyttes til deg uten at man har denne kodelisten, som kun leder for studien og PhD-kandidaten har tilgang til. All informasjon blir altså lagret sikkert.

Vi har taushetsplikt og alle svarene blir behandlet konfidensielt. Det betyr at ingen andre får vite hva akkurat du svarer på spørsmålene vi stiller. Du kan når som helst få innsyn i hvilke opplysninger som er registrert om deg. Du har rett til å få endret på eventuelle feil i de opplysningene vi har registrert. Du har også rett til å få innsyn i sikkerhetstiltakene ved behandling av opplysningene.

For å kunne øke kunnskap om dine og andres opplevelser, og hjelpe med å bedre oppfølgingen, vil vi formidle dette i artikler til internasjonale tidsskrifter, og som en del av et doktorgradsarbeid. I transkripsjoner og publikasjoner vil det ikke være mulig å gjenkjenne deltakerne.

Prosjektleder, lege, Anne Wæhre (tlf: 23 07 18 16), har ansvar for at opplysninger om deg blir behandlet på en sikker måte. Informasjon om deg vil bli anonymisert eller slettet senest fem år etter prosjektslutt.

FORSIKRING

Alle som deltar i forskningsprosjekter gjennom OUS dekkes av pasientskadeloven (lov om erstatning ved pasientskader).

GODKJENNING

Studien er godkjent av Regional komite for medisinsk og helsefaglig forskningsetikk (referanse nr. 79444) og OUS personvernombudet.

Etter ny personopplysningslov har behandlingsansvarlig Oslo Universitetssykehus og prosjektleder Anne Wæhre et selvstendig ansvar for å sikre at behandlingen av dine opplysninger har et lovlig grunnlag. Dette prosjektet har rettslig grunnlag i EUs personvernforordning artikkel 6 nr. 1a og artikkel 9 nr. 2a og ditt samtykke.

Du har rett til å klage på behandlingen av dine opplysninger til Datatilsynet.

KONTAKTOPPLYSNINGER

Dersom du har spørsmål til prosjektet kan du ta kontakt med:

Line Mediå, telefon: 23 07 53 64 /23 07 53 40, E-post: Lmedia@ous-hf.no

Personvernombud ved institusjonen er Tor Åsmund Martinsen. E-post: personvern@oslo-universitetssykehus.no

SAMTYKKE TIL DELTAGELSE

Dersom du ønsker å delta i studien og/eller samtykker i at foreldrene dine får invitasjon til å delta i studien, krysser du av i aktuelle bokser, signerer og sender denne siden til oss i vedlagt frankert konvolutt.

Ja, jeg ønsker å delta i studien ☐

Ja, jeg samtykker i at mine foreldre kan få invitasjon til å delta i studien ☐

Sted og dato

Deltakers signatur

Deltakers navn med trykte bokstaver

Kontaktinformasjon:

Telefon: ……..

Adresse:……
.

Foreldres navn:……

Telefonnummer:……

.

Appendix 6: Information sheet and consent form Healthcare professionals

Senter for Sjeldne Diagnoser
Postboks 4950 Nydalen
0424 Oslo
Sentralbord: 23 07 53 40

Forespørsel om deltakelse i forskningsprosjekt for fagpersoner:

Livssituasjon for personer med variasjon i kroppslig kjønnsutvikling i Norge

Bakgrunn og hensikt

I norske myndigheters handlingsplan mot diskriminering på grunn av seksuell orientering, kjønnsidentitet og kjønnsuttrykk, 2017-2020 – «Trygghet, mangfold, åpenhet», står det at det er viktig å få en bredere kunnskapsbase om personer med variasjon i kroppslig kjønnsutvikling og deres behov for helsetjenester. Et av målene fremhevet i handlingsplanen er å utvikle forskningsbasert kunnskap om livssituasjonen til personer født med variasjon i kroppslig kjønnsutvikling basert på en kvalitativ studie om levekår og utfordringer.

Denne studien er et doktorgradsprosjekt som har utspring i resultatene fra studien gjort på oppdrag fra Barne-, ungdoms- og familiedirektoratet (Bufdir) gjennomført av Senter for sjeldne diagnoser (SSD), Oslo Universitetssykehus (OUS) i 2018. Doktorgraden har til hensikt å gi kunnskap om sårbare perioder inkludert overgang fra barn til voksen, livssituasjon, perspektiver på kirurgi, behov og utfordringer blant personer med variasjoner i kroppslig kjønnsutvikling og deres foreldre. Kvalitative tilnærminger er utforskende og velegnet til å lære fra personer som selv lever med tilstanden, og deres pårørende, om deres egne opplevelser og perspektiv. Deltakernes refleksjoner og opplevelser vil være verdifulle bidrag til å øke kunnskapen om hvordan disse personene kan ivaretas på best mulig måte.

Deltakere vil inkludere ungdom og unge voksne (fra 16 år - 26 år) født med variasjoner i kroppslig kjønnsutvikling, samt foreldre til den samme gruppen. I tillegg ønsker vi å snakke med fagpersoner for å få belyst deres perspektiv.

Appendix 6: Information sheet and consent form Healthcare professionals

Hva innebærer studien?

Du inviteres til å delta på et fokusgruppeintervju (FGI) sammen med flere andre fagpersoner som har erfaring fra å jobbe med personer med DSD. Intervjuet vil finne sted etter nærmere avtale, og tar omtrent 1-1.5 time. Intervjuet blir tatt opp, og transkribert ordrett. Intervjuet vil foregå i gruppe av 6-8 personer som alle har direkte tilknytning til en av de to Flerregionale behandlingstjenestene for usikker somatisk kjønnsutvikling på Haukeland Universitetssjukehus eller OUS.

Mulige fordeler og ulemper

Fordelene med å delta er at du får anledning til å fremme dine tanker og refleksjoner om arbeidet med denne gruppen, samt tanker om eventuelle endringer og forbedringer. FGI gir også en spennende mulighet til å drøfte spørsmål som dukker opp i en klinisk hverdag og høre andre fagpersoner med kjennskap til pasientgruppens refleksjoner og tanker. Mulig ulempe er tiden intervjuet tar.

Hva skjer med informasjonen om deg?

Informasjonen som skal brukes i forskningsprosjektet skal kun brukes slik som beskrevet i hensikten med studien. Alle disse opplysningene vil bli behandlet uten navn eller andre direkte gjenkjennende opplysninger. En kode knytter deg til dine opplysninger og det er kun prosjektmedarbeidere som har adgang til kodelister. Vi vil gjøre vårt ytterste for at andre ikke kan identifisere deg i resultatene av studien når disse publiseres. For eksempel vil ikke profesjon, spesialitet, kjønn eller alder bli knyttet til opplysninger fra gruppeintervjuet, heller ikke hvor eller i hvilken omstendighet gruppeintervjuet er gjennomført.

Frivillig deltakelse

Det er frivillig å delta i studien. Du kan når som helst og uten å oppgi grunn trekke ditt samtykke til å delta i studien. Dersom du ønsker å delta i forskningsprosjektet, undertegner du den vedlagte samtykkeerklæringen. Dersom du trekker deg fra studien, kan du kreve å få slettet innsamlede opplysninger, med mindre opplysningene allerede er inngått i analyser eller brukt i vitenskapelige publikasjoner. Studien er godkjent av Regional komite for medisinsk og helsefaglig forskningsetikk, (referansenummer 79444) og OUS personvernombudet

Dersom du har spørsmål til prosjektet kan du ta kontakt med:
Line Mediå, Telefon: 23 07 53 64 / 23 07 53 40, E-post: Lmedia@ous-hf.no

Personvernombud ved institusjonen er Tor Åsmund Martinsen, personvern@oslo-universitetssykehus.no

Appendix 6: Information sheet and consent form Healthcare professionals

Samtykke til deltakelse i forskningsprosjekt:

Livssituasjon for personer med variasjon i kroppslig kjønnsutvikling i Norge

Jeg har mottatt informasjon om studien og er villig til å delta i fokusgruppeintervju

…… ……

Navn Dato

……

Underskrift

Appendix 7. Errata list

Navn kandidat: Line Mediå

Avhandlingens tittel: LIVING WITH DIFFERENCES IN SEX DEVELOPMENT/INTERSEX. Disclosure, sexual health, perspectives on surgery and stigma

Dokument	Side	Linje	Originaltekst	Korrigert tekst
Kappe del 1- korreksjon	1- Tittelside	1	LIVING WITH DIFFERENCES IN SEX DEVELOPMENY	LIVING WITH DIFFERENCES IN SEX DEVELOPMENT*/INTERSEX*
	8	6	e	e*t*
	8	13	DSD	DSD*/intersex*
	8	30	Vi lærte at for voksne i alderen	Vi lærte at ~~for~~ voksne i alderen
	10	1	HCP	H~~C~~P
Kappe del 1 - korreksjon	2	10	received	receive~~d~~
	6	21	Male hermaphroditism	Male *pseudo*hermaphroditism
	23	8	winder	wi~~n~~der