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Psychosocial and psychosexual aspects of disorders of sex development

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Psychosocial aspects of the treatment of disorders of sex development (DSDs) concern gender assignment, information management and communication, timing of medical interventions, consequences of surgery, and sexuality. Although outcome is often satisfactory, a variety of medical and psychosocial factors may jeopardise the psychological development of children with DSDs. This sometimes results in the desire to change gender later in life. The clinical management of gender dysphoria in individuals with DSD may profit from methods and insights that have been developed for gender dysphoric individuals without DSD. In DSD care, clinical decisions are often made with long-lasting effects on quality of life and should be based on empirical evidence. Yet, such evidence (e.g., regarding gender assignment, information management and timing of surgery) is largely non-existent. DSD-specific protocols and educational materials need to be developed to standardise and evaluate interventions in order to facilitate decision making of professionals and individuals with DSD and enhance psychosocial care in this area.

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Introduction

The treatment of individuals with disorders of sex development (DSDs) requires the input of many disciplines. Although the medical–surgical treatment is highly important, the quality of life also largely depends on the psychosocial management. The new ESPE/LWPES DSD guidelines¹ indeed acknowledge the significance of psychosocial care and the involvement of mental health staff with expertise in DSD.

Several psychological and psychosexual issues that are relevant in DSD care, such as gender identity development, information management, psychological aspects of surgery, sexuality and the

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management of gender dysphoria (see below for definitions) and gender reassignment are addressed in this article. Although there are differences in the care of DSD and non-DSD persons with gender dysphoria, some aspects are strikingly similar.² Clinicians involved in the treatment of DSD may profit from the knowledge and experience in the area of non-DSD gender dysphoria. Therefore, a description of the key issues in the clinical management of non-DSD gender dysphoric conditions is provided as well.

Definitions

In the field of DSD, it is important to make a distinction between the terms 'gender identity' and 'gender role'. The concept of gender identity encompasses cognitive and affective components. For long, cognitive–developmental psychologists have largely focussed on cognitive components of gender identity alone,³ and much of the extensive research in this field concentrates on the implications of achieving more or less sophisticated levels of understanding of being a boy or a girl. Affective components, however, such as feelings of contentment with one's gender⁴ are increasingly included in their conceptualisation of gender identity, which brings their work on gender identity much closer to the clinical field. The term gender identity is mostly used by clinicians to denote a persons' sense of themselves as being male, female or indeterminate.⁵

Gender role refers to behaviours, attitudes and personality traits that a society, in a given culture and historical period, designates as masculine or feminine, that is, more typical of the male or female social role.⁶ In young children, gender role behaviour includes phenomena such as preference for same-sex versus other-sex peers, roles in fantasy play, toy interests, dress-up play and interest in rough-and-tumble play. In older children or adults, gender role is measured using personality attributes with stereotypic masculine or feminine connotations or with regard to recreational and occupational interests and aspirations.⁶

The term transsexualism refers to a desire for complete gender reassignment originating from an experienced discrepancy between one's gender of assignment, on the one hand, and one's basic sense of self as a male or female, or indeterminate (gender identity), on the other.

In 1973, Fisk⁷ proposed the term gender dysphoria syndrome. This term allows for the possibility that an atypical gender identity development does not always result in a complete cross-gender identity and wish for gender reassignment. Gender dysphoria is now often used to indicate the distress associated with conflicting gender identity and gender of assignment. A discrepancy between assigned gender and gender identity and/or role, however, does not always seem to be associated with distress.⁸ For this reason, more neutral terms such as gender (identity) variance, gender dissonance, gender incongruence or transgenderism are often used² for the spectrum of atypical manifestations of gender identity and gender role phenomena. Although in the current version of the psychiatric classification system of the American Psychiatric Association, *The Diagnostic and Statistical Manual of Mental Disorder*,⁹ the term Transsexualism was replaced by Gender Identity Disorder (GID), the former term is still often used. Strong cross-gender identification and an extreme aversion to one's assigned sex and gender role are the core aspects of both transsexualism and GID.

Gender assignment

Decisions on gender assignment can only be made after a thoughtful balancing of medical and psychosocial pros and cons. Gender assignment will depend on the estimated optimal outcome. Unfortunately, in some conditions, adult gender identity cannot easily be predicted.^{10,11} This is because, still, little is known about the relative contribution of biological (e.g., genes and prenatal sex hormone exposure) and non-biological influences (e.g., parental attitude, peer influences and cultural context) on gender identity development. Critical phases of development or potential interactions of the various factors are also unclear.

The most extensively studied condition with regard to psychosexual outcome is congenital adrenal hyperplasia (CAH). Women with CAH, who were raised in the female gender, mostly have female gender identities. However, a less strong female identification, gender discomfort and even gender dysphoria occur considerably more often in this group than in women without DSD.^{12,13} Reviewing

studies on psychosexual outcome in DSD up to 2007, de Vries et al.¹³ found that 10 of 217 (5%) of adolescents raised as female and adult women with CAH had some form of gender dysphoria. This sometimes even resulted in a gender change. Male gender role behaviour appeared to be common. Girls with CAH have more masculine and less feminine interests regarding toys, clothing and make-up, infant care, sports and playmates. Most of the findings on gender role behaviour that are suggestive of an effect of prenatal brain exposure to androgens have been found to be stronger in the more seriously affected salt-wasting than in the simple virilising group.¹⁴

More dramatic is the gender identity outcome in 5 α -RD-2 and 17 β -HSD-3. When raised as boys, these children have a male identity and behave like boys. However, when such children are raised as girls, the outcome is more varied. After the first observations in the Dominican Republic of gender changes in 5 α -RD-2 individuals raised as female,¹⁵ gender changes were reported in 17 β -HSD-3 and from other societies as well. Such changes, however, did not happen in all affected individuals, even when they were living in societies that highly value the male role. Unfortunately, virtually no reports give any detailed information on parental rearing styles, psychosexual development or more subtle signs of gender discomfort. Such information is crucial for the understanding of the apparent variation in gender identity outcome. In the review by de Vries et al.,¹³ 69 of 117 female-raised 5 α -RD-2 individuals (59%) and 20 of 51 17 β -HSD-3 individuals (39%), all older than 12 years, had gender dysphoria to the extent that they decided to live as males. In one study by Richter-Appelt, Discher and Gedrose,¹⁶ five female-raised persons with 5 α -RD-2 and 17 β -HSD-3 had a significantly lower 'female gender identity' score than female controls on a scale measuring this concept. One of the five also reached a significantly higher male gender identity score than female controls on a male gender identity scale. Whether this signifies clinically significant gender dysphoria is not clear.

Individuals with complete androgen insensitivity syndrome (CAIS), raised as girls, have a female gender identity. They usually are described as very feminine in their gender role behaviour, but there may be more variability in their behaviour than has long been assumed. In a comparison between women with CAIS recruited through the United Kingdom Database of Ambiguous Genitalia and Intersex Disorder and an Androgen Insensitivity Support Group, it appeared that the first group recalled more masculine-typical toy, activity and playmate preferences as children and overall ratings of childhood masculinity than the support group.¹⁷ Furthermore, in the Richter-Appelt et al. study, two of five women with CAIS scored low on female gender identity.¹⁶ However, in the review by de Vries et al.,¹³ not one of 98 women with CAIS reported to suffer from gender dysphoria or made a gender change. The picture is different, however, in partial androgen insensitivity syndrome (PAIS). In 46 female-raised adolescent and adult individuals, five (11%) were gender dysphoric or changed gender. This percentage was even higher in the male-raised group, as five of 35 (14%) was gender dysphoric or changed gender. All gender changes took place in adulthood. In the Richter-Appelt et al. study,¹⁶ one of three female-reared individuals with PAIS scored significantly lower than female controls on the female gender identity scale.

From these studies and those on conditions such as penile agenesis, penile ablation, cloacal exstrophy of the bladder and micropenis,^{18,19} it appears that testosterone influences the development of a male gender role and seems to increase the chance of a male gender identity. However, in prenatally testosterone-exposed individuals raised as girls, a female adult gender identity is the rule rather than the exception.

When gender assignment decisions have to be made, matters such as surgical possibilities, potential for fertility and need for hormone replacement are included.¹ In these decisions, non-medical factors such as the potential for psychological functioning as male or female in terms of gender role and sexuality and the ability of parents to cope with uncertainties and complexities related to one decision or the other have to be taken into account as well.²⁰ Evidence on the functioning and quality of life of adults with DSD that could be used to facilitate such decisions is currently scarce, but, fortunately, the number of follow-up studies, paying appropriate attention to quality of life and psychosexual functioning, is increasing.²¹

Most parents find it highly traumatic if the sex of their newborn cannot immediately be determined. Even after the diagnosis is made, the parents' ability to cope with uncertainties on the child's development may be tested. Mental health professionals need to estimate to what extent parents are able to

cope emotionally with the complexities of their child's condition. In this estimation, parental, religious and cultural factors have to be taken into consideration. Besides parents' emotional coping abilities, their capacities to handle practical matters and their understanding of their child's condition are critical in gender assignment decisions. Screening tools for identifying parents at risk for maladaptive coping to children's illnesses do exist,²² but more DSD-specific tools are needed. It would be very helpful if such material would be easily obtainable, for instance, on the Internet.

Especially if families are already vulnerable for psychological distress, educational sessions alone may not suffice. To avoid potential harm of the child by the parents' escalating distress, psychologists may need to train them in certain problem-solving skills. However, DSD-specific programmes hardly exist. The challenge for mental health professionals is to realistically inform the parents about potential concerns during the child's development without making them unnecessarily anxious.

Information management

Information management involves disclosure of information both from clinicians to parents and child and from child/family to the wider environment. The most sensitive information concerns karyotype and gonadal status, especially when these aspects are not concordant with the gender of assignment and infertility. Nevertheless, children also deserve explanations for other matters they encounter on a regular base. For instance, some should know why they need certain types of medication in childhood, or hormone replacement at puberty, why they have surgical scars or for what their regular medical check-ups are meant. Others have to dilate their vaginas or have to make decisions about surgery. Information management requires more than presenting information comprehensibly, because the information may raise difficult and emotional issues. For instance, atypical gender role behaviours and sexual feelings (sexual attraction to same-gender peers) may create anxiousness if the condition is not fully understood, and wrong conclusions are drawn regarding one's 'true' sex. Because of the emotional load, cognitive processing may be less than optimal. Over the years, parents and children often need repeated explanations. The information always has to be adapted to the parents' capacities to understand and the child's developmental stage. Most clinicians would agree that the children should be fully informed by the time they reach adulthood, and are no longer treated by their paediatricians but monitored by endocrinologists, gynaecologists or other specialists.

The DSD consensus document¹ recommends early planning with the parents and recurrent, gradual medical education and counselling. This approach is a sensible one, but, in practice, it is sometimes difficult to deal with resistance of parents to inform the child. Parents fear that this information may deprive their children of a happy childhood or create emotional and relational problems. If cultural factors play a role, disclosure may be even harder to deal with, as conceptualisations and values regarding gender and sexuality differ largely between cultures.²³ Yet, from other areas in medicine, it is known that being informed about one's medical condition is associated with psychological adjustment.²⁴ Timely and well-informed children will have better opportunities to develop coping skills and will have a better chance to develop a more positive self-image and incorporate limitations such as infertility in their expectations about their future lives. The ability of children to fully understand relevant aspects of their condition and give proper informed consent is especially important when they have to decide on medical interventions.

Counselling the family also implies discussing how to deal with the social environment. It is still an open question whether sharing information with the wider environment will have positive or negative consequences for the child. Is living a 'normal' life with a secret more harmful than living a life without secrets but with a reasonable chance for stigma or shame? The advices that are now given to parents are still largely based on the clinicians' personal opinions what would be favourable, considering their evaluation of the family, the child and their wider social and cultural environment.

To improve our care with regard to information management, we would need to have better insight into the effects of timing, type of information (and a potential interaction of the two), the best way of conveying information and into the influence of cultural, family and child factors on how the information is dealt with. How the influence of these factors changes over time (e.g., due to characteristics of

the condition) is also unexplored. Studies comparing the development of children who actually lived in 'informed' versus 'uninformed' environments would be of great use. Such studies could form the base of protocols on information management. The information, however, should always be adapted to the individual's capabilities.

Psychosocial aspects of surgery

The DSD consensus document¹ offers guidelines on genital surgery (e.g., in case of severe virilisation in female-assigned children; gonadectomy in female-raised PAIS and CAIS children). Yet there are still choices to make for older children and parents, for instance, regarding the timing of surgery and vaginal dilatation. They have to be prepared for such decisions. Because, in some cases, there is not enough evidence to support one or the other choice, and certain choices (e.g., timing of vaginal dilatation in early or later adolescence) very much depend on individual characteristics of the person, the advantages and disadvantages of the various choices have to be discussed. In particular, sexual consequences and goals of genital surgery should be addressed.

Educational materials, developed by children's hospitals or support groups, are easily available on the Internet. Some can be used for this purpose, but the information is not always of high quality and can even be confusing without individualised counselling. Systematic decision-making aids, as developed for some conditions (see The Foundation for Informed Medical Decision Making), would be very valuable. Although many families highly appreciate support group contact, some avoid any confrontation with other persons with DSD. Whether support group contact actually contributes to surgery decisions has never been studied systematically.

Children who undergo surgery have to be prepared. Behaviour therapy techniques for other conditions have been found to be beneficial,²⁵ but have to be adapted to DSD.

Sexuality

In adolescence, sexuality and pair-bonding become important aspects of life. Adolescents with DSD may find it difficult to enter this phase of life without anxieties, especially if they have a history of repeated genital exams and medical photography, disrespectful treatment by clinicians or have an atypical genital appearance. After entering puberty, some feel increasingly uncertain about their masculinity/femininity, sexual adequacy or sexual orientation. They often postpone initiating intimate relationships because of their insecurities and fear of rejection. Sexual problems indeed occur more often in DSD than in non-DSD groups.^{26,27} For instance, the sexual lives of women with CAH differ from control groups in terms of timing of psychosexual milestones (delayed), sexual experiences (less), sexual activity and imagery (less), sexual motivation (less), partnership and marriage (less) and sexual self-image (less favourable).^{28–30} Most women with CAH are heterosexual, but the incidence of homosexuality as compared to population norms or other control groups is increased.^{30,31} Very little is known about the sexual functioning and sexual orientation in individuals with enzyme deficiencies. Regarding the persons who were raised as girls, there are occasional referrals to marriage and sexual intercourse, heterosexual attraction and relationships with men.³² Various sexual problems and an elevated percentage of non-heterosexuality were recently reported in seven women with enzyme deficiencies,²⁷ but systematic studies in larger groups are lacking. In the same study, sexual problems, primarily low sexual desire, and inability to become sexually aroused and have orgasms have also been reported by 10 women with CAIS.²⁷ The sexual orientation of women with CAIS is heterosexual.^{27,33–35} In a study comparing women with CAIS recruited through the United Kingdom Database and the Support Group, the Database group appeared to be more strongly heterosexual, but, in general, the women with CAIS did not differ from other women with regard to marriage and relationship patterns.¹⁷ In a small group of women with PAIS, most feared to have sexual contacts, had experienced dyspareunia or fear of becoming hurt by sexual contacts. There was also an increase of non-heterosexuality.²⁷

Because of the elevated risk of sexuality-related problems, an adequate and timely preparation regarding sexuality and relationships is of great importance. Adolescents do not only need the necessary medical and sexual education, but they should also have the opportunity to discuss their concerns repeatedly and in private with a mental health clinician.

Gender reassignment

If a correct diagnosis is only made after the child has been assigned to one gender, but the diagnosis suggests that it would be more appropriate to assign the child to the other gender, the question arises until what age a physician-imposed gender change can take place without harming the child's development. Developmental psychology studies⁶ show that most children can correctly label their gender at about 30 months, some even earlier, but when exactly affective aspects of gender identity develop and until what age these aspects remain flexible is unclear. Furthermore, it is likely that, in children with DSD, the process of gender development occurs differently from the typical gender development. The often mentioned age limit of 18 months is still much debated. However, even for children under 30 months, it seems wise to be increasingly careful with imposed gender change when the child grows older.

When the child grows up, it is helpful for parents to have regular contacts with the team, as they may struggle with issues that they, correctly or not, attribute to the child's DSD. A frequent concern of parents regards gender role behaviour that is inconsistent with gender of assignment and rearing. They often take this as an indication of gender dysphoria or even a wish for gender reassignment. Parents may fear that the gender assignment decision at birth had been wrong, and this may make them feel guilty.

If there are persistent and strong indications that gender dysphoria is present, a comprehensive assessment by clinicians, skilled in the management of gender change, is essential, irrespective of the age of the person. Only after thorough assessment, steps in the direction of gender reassignment should be taken. Clinicians dealing with gender change in individuals with DSD may profit from the insights, which have been developed in the area of gender reassignment in non-DSD gender dysphoria. To assist clinicians without specific expertise in gender dysphoria, the current guidelines for gender reassignment in non-DSD individuals (the Standards of Care of the World Professional Association of Transgender Health or WPATH, a professional organisation in the field of gender dysphoria)³⁶ could be adjusted or expanded and more specifically address the care for gender-dysphoric individuals with DSD.² Below is a description in what way clinical management of non-DSD gender-dysphoric persons may be used for DSD individuals with gender dysphoria.

The prevalence of extreme forms of gender dysphoria in children and adolescents is not known. In adults, the DSM-IV-TR reports prevalence rates of extreme forms of gender dysphoria (GID or transsexualism) of 1:30 000 in males to 1:100 000 in females,⁹ but much higher rates have been reported in various countries.³⁷

The clinical management of gender dysphoria in individuals without DSD largely depends on the age of the referred person. In children, the clinician assesses whether they meet the DSM-IV-TR core criteria⁹ for GID (a strong and persistent cross-gender identification and a persistent discomfort with his or her sex or sense of inappropriateness in the gender role of that sex) and whether clinical intervention is needed. Especially in children, gender-variant behaviour may be more upsetting to the environment than to the child itself, and make the parents rather than the child seek clinical help. The clinician also has to understand what factors (in the past or at the time of referral) may have influenced the gender-variant behaviour and preferences of the child. The information about the child's gender behaviour and feelings then has to be appraised against a broader background, such as general aspects of the functioning of child and family. A proper understanding of the gender problem, evaluated in this wider perspective is necessary when a choice about appropriate interventions has to be made. A number of instruments are available to assess gender understanding, behaviour and feelings of children.³⁸ These are cognitive tests, parent and child questionnaires, child interviews, play observations and projective methods.

Although there is disagreement³⁹ regarding the psychological treatment of gender-variant children, because their treatment is considered by some as an effort to prevent later homosexuality, most clinics offer some form of psychological treatment. These interventions are sometimes aimed at diminishing the gender-variant behaviour itself, but more often at helping parents to maximise opportunities for their children's adjustments.^{40,41} Nearly all these diagnostic tools and procedures, as well as the psychological interventions used, are easily applicable in cases of gender-dysphoric children with DSD.

In adolescents and adults without DSD who apply for gender reassignment, the gender problem is also examined comprehensively in the diagnostic phase. Because of the importance of the decision that has to be made, the diagnostic procedure is rather extensive. The recommended procedure in the Standards of

Care of the WPATH³⁶ is to come to the gender-reassignment decision in steps. In the first diagnostic phase, it has to be established that an applicant fulfils DSM or ICD criteria for the diagnoses of GID or transsexualism. The next phase includes three elements, labelled triadic therapy, for those who are eligible for gender reassignment. The elements consist of a 'real-life experience' (RLE) in the desired role, hormones of the desired gender and surgery to change the genitals and other sex-related characteristics.

In the first, diagnostic, phase, information is obtained from the gender-reassignment applicant and, in case of adolescents, the parents on the general and psychosexual development of the applicant. Information is gathered about current cross-gender feelings and behaviour and current social, sexual and psychological functioning. In order to prevent unrealistically high expectations as regards their future lives, the applicants also have to be thoroughly informed about the possibilities and limitations of gender reassignment and other kinds of treatment. In case of DSD applicants for gender reassignment, specific attention has to be paid to surgical limitations because of previous operations. Those who are merely gender-confused or have a wish for gender reassignment that seems to originate from factors other than a genuine and complete cross-gender identity are served best by psychological interventions alone and are usually referred to psychotherapists. Such interventions may help them to better understand and cope with gender issues, and to try out alternative solutions to their problem, such as part-time cross-gender living. However, applicants who are eligible for gender reassignment may also profit from psychotherapy, because they need time to reflect on unresolved personal issues or doubts regarding gender reassignment before they embark on somatic treatment. Other issues that may come up are anxieties concerning the loss of family or friends, uncertainties whether passing in the other gender role is feasible, and concerns on the possibility of having satisfying intimate relationships and support when 'coming out'. These issues are also typically addressed when DSD persons who are gender dysphoric and consider changing gender are counselled.

Those who are eligible for gender reassignment start with the RLE phase. In this phase, one has to live full time in the desired role. The RLE is defined as "the act of fully adopting a new or evolving gender role or gender presentation in everyday life," with the intention of experiencing *in vivo* the familial, interpersonal, socioeconomic and legal consequences of transition.³⁶ The RLE is meant to test the person's resolve, the capacity to function in the preferred gender, and the adequacy of social, economic and psychological supports. A fundamental premise of the RLE is that the person should experience life in the desired role before making irreversible physical changes. The RLE includes informing family, friends, work, school and other social contacts about the intention to undergo gender transition. Usually, a new name congruent with the person's gender identity is chosen, and there is a concurrent switch in gender pronouns. There may also be a change in clothing, hairstyle and gender-specific behaviours (e.g. use of the men's washroom for female-to-males). During the RLE, the person's feelings about the social transformation, including coping with the responses of others, is a major focus of the discussions and counselling. A period of testing living in the desired gender role would also be of great value for gender-dysphoric individuals with DSD who intend to change gender.

Physical intervention (i.e., hormonal and/or surgical feminisation/masculinisation) is a long-term process. The idea is that treatment should thoughtfully and recurrently be considered over time. Informed consent is essential, as it is for any type of physical intervention. The different steps in physical interventions and the duration of the process require that some information is repeatedly given and discussed. Guidelines of the Royal College of Psychiatrists⁴² and the WPATH Standards of Care³⁶ distinguish between fully reversible, partially reversible and irreversible stages of physical interventions for adolescents. For adults, the fully reversible interventions do not apply.

Different views exist on at what age and at what pubertal stage to start with puberty-delaying hormones. To let adolescents experience their physical puberty at least to some extent, some clinicians choose to wait until Tanner stage 2 or 3, whereas others believe it is better to wait even longer. In addition to standard readiness criteria relating to psychologic stability sufficient to withstand the stresses of gender reassignment, a number of additional criteria must be met for pubertal delay, such as an early-onset age of the gender dysphoria, consistency in gender dysphoria around puberty and consenting and participating parents. Guidelines for the endocrine treatment of transsexuals have recently been published by the Endocrine Society.⁴³ Typically, GnRH analogues such as leuprolide or triptorelin are used to delay/suppress puberty. In contrast to gender-dysphoric adolescents without DSD, agonadal adolescents with DSD do not need suppression of puberty. They may, however,

sometimes profit from some delay in the start of hormone replacement therapy. Like in their counterparts without DSD, this may give them more time to reflect on their wish to change gender and allow for some time to prepare themselves psychologically and practically for the gender change. As there is so much variation in the types and degrees of DSD conditions, these gender changes should always be tailored to the individual needs.

Feminising/masculinising hormone therapy (i.e., oestrogens/anti-androgens/progestins for male-to-females and androgens for female-to-males) is considered partially reversible, as some of the changes persist even if hormone therapy is discontinued. Some changes (e.g., breast growth in male-to-females or facial hair growth in female-to-males) require surgery or other treatment to 'reverse'. Because of the risks involved in feminising/masculinising hormone therapy, this typically does not begin until the applicant is 16 years or older. From a paediatric endocrinologist's point of view, it is argued that, irrespective of the Tanner stage at presentation for treatment, adolescents undergoing gender reassignment should be treated with GnRH analogues first to keep their own sex hormone production low. Androgens or oestrogens are then gradually added to induce puberty of the desired sex. Because of the differences between DSD and non-DSD conditions, age limits for the start of these hormones do not need to be the same for adolescents with DSD.

Gender reassignment surgery is not carried out prior to adulthood in young gender dysphorics without DSD. There is international clinical consensus that the risks of early surgical intervention far outweigh the potential benefits in virtually all cases. The surgical feminisation/masculinisation is explained in detail in a document 'Care of the Patient Undergoing Sex Reassignment Surgery'.⁴⁴ Again, considering the differences between the conditions, this limit is not applicable to adolescents with DSD.

Summary

Although the psychological outcome of DSD is often satisfactory, medical and psychosocial factors may put the development of children with DSD in many ways at risk. Even in adulthood, certain aspects of life may remain challenging. Gender assignment or physician-imposed gender reassignment may result in the wish to change gender by the person involved; genital ambiguity may cause parental rejection; or genital surgery may create sexual dysfunctions, to name a few. Besides the DSD, clinical decisions, the timing of interventions and the way medical information is communicated have far-reaching consequences for one's quality of life. The consensus document¹ offers guidelines in the clinical management of DSD. However, many aspects of counselling DSD children and their parents are not yet supported by good evidence. Screening tools, DSD-specific (psychological) treatment protocols and educational materials are lacking. The rarity of most DSD conditions often made it difficult to conduct studies addressing many of the above topics. In recent years, international collaboration in the field of DSD is increasing, although so far most of the studies focus on medical aspects. If psychological and social aspects would also be included in such collaborative studies, mental health professionals no longer have to base their work merely on clinical intuition.

Practice points

- Male gender role behaviour in female-raised children should not be mistaken for a male gender identity
- DSD management always needs a multidisciplinary approach; a mental health professional should be part of the team
- DSD team members should avoid giving contradictory information to parents of children with DSD and older individuals with DSD
- Clinicians should timely prepare young adolescents for the consequences of their DSD for their sexual life
- Clinicians should pay specific attention to potential sexual problems in adults with DSD
- Many aspects of the clinical management of gender change that have been developed for individuals without DSD can be used for gender-dysphoric individuals with DSD

Research agenda

- Follow-up studies should appropriately measure quality of life and psychosexual functioning and outcome
- Tools to identify at-risk families and training programmes for at-risk families need to be developed and made easily available, preferably through the Internet
- Practice guidelines should be developed and evaluated for DSD individuals with gender dysphoria or the desire to change gender
- More studies are needed to investigate the effects of timing, type of information, method of conveying information and the role of cultural, family and child factors in information management to enable the development of evidence-based information management protocols
- Educational materials/decision aids should be developed to standardise information management and enable systematic evaluations
- To prepare families properly for genital surgery, educational material and psychological treatment techniques addressing DSD aspects should be developed

References

- *1. Hughes IA, Houk C, Ahmend SF et al. Consensus statement on management of interest disorders. *Archives of Disease in Childhood* 2006; **91**: 554–563.
- *2. Meyer-Bahlburg HFL. Variants of gender differentiation in somatic disorders of sex development. *Int J Transgenderism*, in press.
3. Martin CL, Ruble DN & Szkrybalo J. Cognitive theories of early gender development. *Psychological Bulletin* 2002; **128**: 903–933.
4. Egan SK & Perry DG. Gender identity: a multidimensional analysis with implications for psychosocial adjustment. *Developmental Psychology* 2001; **37**: 451–463.
5. Money J. Lovemaps: Clinical concepts of sexual/erotic health and pathology, paraphilia, and gender transposition in childhood, adolescence, and maturity. New York: Irvington, 1986.
6. Ruble DN, Martin CL & Berenbaum SA. Gender development. In Damon W, Lerner RM & Eisenberg N (eds.). *Handbook of child psychology. Social, emotional, and personality development*. 6th ed 2006; vol. 3. New York: Wiley, 2006, pp. 858–932.
7. Fisk NM. Gender dysphoria syndrome: the conceptualization that liberalizes indications for total gender reorientation and implies a broadly based multi-dimensional rehabilitative program. *The Western Journal of Medicine* 1974; **120**: 386–391.
- *8. Bockting WO. Psychotherapy and the real-life experience: from gender dichotomy to gender diversity. *Sexologies* 2008; **17**: 211–224.
9. American Psychiatric Association. *Diagnostic and statistical manual of mental disorders*. 4th ed. Washington: Author, 2000.
- *10. Berenbaum SA. Psychological outcome in children with disorders of sex development: implications for treatment and understanding typical development. *Annual Review of Sex Research* 2006; **17**: 1–38.
- *11. Cohen-Kettenis PT. Psychological long-term outcome in intersex conditions. *Hormone Research* 2005; **64**: 27–30.
12. Dessens AB, Slijper FME & Drop SLS. Gender dysphoria and gender change in chromosomal females with congenital adrenal hyperplasia. *Archives of Sexual Behavior* 2005; **34**: 389–397.
- *13. de Vries ALC, Doreleijers THAH & Cohen-Kettenis PT. Disorders of sex development and gender identity outcome in adolescence and adulthood: understanding gender identity development and its clinical implications. *Pediatric Endocrinology Reviews* 2007; **4**: 343–351.
14. Meyer-Bahlburg HFL, Dolezal C, Baker SW et al. Gender development in women with congenital adrenal hyperplasia as a function of disorder severity. *Archives of Sexual Behavior* 2006; **35**: 667–684.
15. Imperato-McGinley J, Peterson Gautier T & Sturla E. Androgens and the evolution of male-gender identity among male pseudohermaphrodites with 5-alpha-reductase deficiency. *The New England Journal of Medicine* 1979; **300**: 1233–1237.
16. Richter-Appelt H, Discher C & Gedrose B. Gender identity and recalled gender related childhood play-behaviour in adult individuals with different forms of intersexuality. *Anthropologischer Anzeiger* 2005; **63**: 241–256.
17. Hines M, Ahmed SF & Hughes IA. Psychological outcomes and gender-related development in complete androgen insensitivity syndrome. *Archives of Sexual Behavior* 2003; **32**: 93–101.
18. Mazur T. Gender dysphoria and gender change in androgen insensitivity or micropenis. *Archives of Sexual Behavior* 2005; **34**: 411–421.
19. Meyer-Bahlburg HFL. Gender identity outcome in female-raised 46, XY persons with penile agenesis, cloacal exstrophy of the bladder, or penile ablation. *Archives of Sexual Behavior* 2005; **34**: 423–438.
- *20. Meyer-Bahlburg HFL. Treatment guidelines for children with disorders of sex development. *Neuropsychiatrie de l'Enfance et de l'Adolescence* 2008; **56**: 345–349.
21. Thyen U & Krege S. Netzwerk DSD/Intersexualität. *Der Urologe. Ausg. A* 2007; **46**: 1304–1306.
22. Kazak AE, Cant MC, Jensen MM et al. Identifying psychosocial risk indicative of subsequent resource use in families of newly diagnosed pediatric oncology patients. *Journal of Clinical Oncology* 2003; **21**: 3220–3322.

23. Lang C & Kuhnle U. Intersexuality and alternative gender categories in Non-Western cultures. *Hormone Research* 2008; **69**: 240–250.
24. Wiener L, Mellins CA, Marchefka S et al. Disclosure of a HIV diagnosis to children: history, current research, and future directions. *Journal of Developmental and Behavioral Pediatrics* 2007; **28**: 155–166.
25. Harbeck-Weber C, Fisher JL & Dittner CA. Promoting coping and enhancing adaptation to illness. In Roberts MC (ed.). *Handbook of pediatric psychology*. 3rd ed. New York, London: The Guilford Press, 2003, pp. 99–118.
26. Ismail I & Creighton S. Surgery for intersex. *Reviews in Gynaecological Practice* 2005; **5**: 57–64.
27. Schönbacher V, Schweize K, Rustige L, Schützmann K, Brunner F, Richter-Appelt H. Sexual Quality of life of individuals with 46, XY disorders of sex development. *The Journal of Sexual Medicine*, in press.
28. Kuhnle U, Bullinger M & Schwarz HP. The quality of life in adult female patients with congenital adrenal hyperplasia: A comprehensive study of the impact of genital malformations and chronic disease on female patients' life. *European Journal of Pediatrics* 1995; **154**: 708–716.
29. Slijper FME, van der Kamp HJ, Brandenbrug H et al. Evaluation of psychosexual development of young women with congenital adrenal hyperplasia: A pilot study. *Journal of Sex Education & Therapy* 1992; **18**: 200–207.
30. Zucker KJ, Bradley SJ, Oliver G et al. Psychosexual development of woman with congenital adrenal hyperplasia. *Hormones and Behavior* 1996; **30**: 300–318.
31. Hines M, Brook C & Conway GS. Androgen and psychosexual development: core gender identity, sexual orientation, and recalled gender role behaviour in women and men with congenital adrenal hyperplasia (CAH). *Journal of Sex Research* 2004; **41**: 75–81.
32. Mendonca BB, Inacio M, Costa EM et al. Male pseudohermaphroditism due to steroid 5-alpha-reductase 2 deficiency: diagnosis, psychological evaluation, and management. *Medicine (Baltimore)* 1996; **7**: 64–76. 1996.
33. Lewis VG & Money J. Gender identity/role: G-I/R Part A: XY androgen-insensitivity syndrome and XX Rokitansky syndrome of vaginal atresia compared. In Dennerstein L & Burrows G (eds.). *Handbook of psychosomatic obstetrics and gynaecology*. Amsterdam: Elsevier Biomedical Press, 1983, pp. 51–60.
34. Masica DN, Ehrhardt AA & Money J. Fetal feminization and female gender identity in the testicular feminizing syndrome of androgen insensitivity. *Archives of Sexual Behavior* 1971; **1**: 131–142.
35. Money J, Ehrhardt AA & Masica DN. Fetal feminization by androgen insensitivity in the testicular feminizing syndrome. Effect on marriage and maternalism. *The Johns Hopkins Medical Journal* 1968; **123**: 105–114.
36. Meyer WJ, Bockting WO, Cohen-Kettenis PT et al. The Harry Benjamin international gender dysphoria association's standards of care for gender identity disorders, sixth version. *Journal of Psychology & Human Sexuality* 2001; **13**: 1–30.
37. Zucker KJ & Lawrence AA. Epidemiology of gender identity disorder. *International Journal of Transgenderism* 2009; **11**: 8–18.
- *38. Zucker KJ. Measurement of psychosexual differentiation. *Archives of Sexual Behavior* 2005; **34**: 375–388.
39. Pleak R. Ethical issues in diagnosing and treating gender-dysphoric children and adolescents. In Rottnek M (ed.). *Sissies & tomboys: gender nonconformity & homosexual childhood*. New York: New York University Press, 1999, pp. 34–51.
40. Menvielle EJ & Tuerk C. A support group for parents of gender non-conforming boys. *Journal of the American Academy of Child and Adolescent Psychiatry* 2002; **41**: 1010–1013.
- *41. Zucker KJ & Cohen-Kettenis PT. Gender identity disorder in children and adolescents. In Rowland DL & Inrocchi L (eds.). *Handbook of sexual and gender identity disorders*. New York: Wiley & Sons, 2008, pp. 376–422.
42. Di Ceglie D. Management and therapeutic aims with children and adolescents with gender identity disorders and their families. In DiCeglie D & Freedman D (eds.). *A stranger in my own body: atypical gender identity*. Development and Mental Health London: Karnac, 1998.
- *43. Hembree WC, Cohen-Kettenis PT, Delemarre-van de Waal HA et al. Endocrine treatment of transsexual persons: an endocrine society clinical practice guideline. *The Journal of Clinical Endocrinology and Metabolism* 2009; **94**: 3132–3154.
44. Bowman C & Goldberg JM. Care of the patient undergoing sex reassignment surgery (SRS) Vancouver, BC, Canada: Vancouver Coastal Health Authority, <http://www.vch.ca/transhealth/resources/library/tcpdocs/guidelines-surgery.pdf>; 2005.