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Modified terms of reference for the Senate Inquiry

The involuntary or coerced sterilisation of people with disabilities in Australia: People with intersex conditions

This submission is from members of the Disorder of Sex Development multidisciplinary team at the Royal Children's Hospital, Melbourne. This clinical service includes clinicians from the following Departments:Paediatric Endocrinology, Paediatric and Adolescent Gynaecology, Paediatric Urology, Social Work, and Bioethics. The individual clinicians involved in the provision of this service have had experience specific to this field ranging from 3 years to more than 30 years.

Modified Terms of Reference for Senate Inquiry on involuntary or coerced sterilisations of people in Australia

Current practices and policies relating to the involuntary or coerced sterilisation of intersex people, including:

(a) sexual health and reproductive issues; and

(b) the impacts on intersex people.

Introduction

In medical nomenclature, intersex conditions are now referred to as Disorders of Sex Development (DSD), following an international consensus meeting involving medical experts and advocates in 2006. We do acknowledge that some affected individuals dislike this term, but as it is the current internationally accepted nomenclature, we have elected to use this term throughout this submission.

The outcomes for some people with DSDs in the past have not been as good as one would hope for by today's standards of care. This includes outcomes relating to mortality, morbidity, and health in its broadest definition. Many of these issues continue for children and people with these conditions around the world today.

This submission will begin by giving an introduction to the range of conditions that are known as DSD. This submission will not replicate the medical details and principles as outlined in the APEG submission. The purpose of this submission is to clarify the changes in practice over the last few decades, to outline the factors that may have contributed to the poor outcomes and problems experienced by adults and to highlight the current efforts to achieve better care and outcomes. Additionally it is important to appreciate the factors that have influenced, and continue to influence the longterm outcomes of people with DSD. A comparison of the past versus the current management approaches for some of these conditions will be outlined, as well as some of the challenges. A detailed description of the full range of DSDs will not be made. Many of these details have been submitted in the APEG submission, which the RCH supports.

Background information

DSDs are a complex set of conditions whereby the chromosomal, hormonal or physical makeup of an individual is atypical. These conditions are challenging – for the individual, their family, as well as from a healthcare management and treatment perspective.

The range of conditions that fulfil the international criteria for intersex or DSD include:

- Some life threatening conditions such as salt wasting congenital adrenal hyperplasia, which requires life long medications and medical care;
- Babies born with ambiguous genitalia;
- others which involve significant penis anomalies (hypospadias);
- others involving girls who are born without a vagina and uterus; and

- babies who are born with only one opening for bladder, bowels (and vagina) or where the entire lower abdominal wall and genital area is open and exposed with the inside of the bladder open and the clitoris or penis in 2 un-joined halves.

Some of these conditions are obvious at birth, others may not be identified until periods fail to start in a teenage girl and others not recognised until infertility (in a man or a woman) is investigated. In some parts of the world some of these conditions continue to result in death of the newborn child or infant.

Some of these conditions require immediate major surgery to close or unblock structures such as bowel and bladder. Some conditions require no surgery at all. Some girls will require genital surgery for outflow of menses and sexual function. Some of these conditions pose significant complex ethical issues.
All children with these conditions and their families will need sensitive and individualised care specific to their condition with appropriate emotional support.

Changes in approaches to management of individuals with DSD over time

As with many facets of society and medicine there have been substantial changes in the care of children and people with DSDs over the last few decades. This relates not only to increased medical knowledge and better surgical techniques, but also different societal attitudes and perspectives. In the past, clinical care and practices which were in keeping with medical and societal attitudes were less sensitive to the psychosocial impact of the diagnosis on the young person and their family. These practices have evolved over time as community and medical attitudes has progressed. From a medical perspective, improved knowledge regarding cancer risks of different DSDs, as well as improved capacity to measure the hormonal production and potential fertility of gonads have been particularly seminal changes. Health care provision needs to encompass sensitivity to the families and the individual.

The changes in care of a young person with a DSD recognise the need to be more sensitive, less intrusive and more supportive of the young person's development of a positive body image and self esteem. Establishing quality of life, with the capacity to enjoy positive intimate relationships, and where possible maximizing fertility potential, are universal treatment goals.

Fertility, cancer risk, sterilizing procedures and DSD

Quite a number of the conditions that are included under the definition of DSDs are associated with an increased cancer risk as well as infertility.

1. XY Complete gonadal dysgenesis. Individuals with this condition may have both the external physical appearances of a girl and a uterus, and will most likely identify as female. However, due to problems present during fetal development, their gonads are neither testes nor ovaries, but rather underdeveloped structures without potential for hormone production or fertility. The term 'streak' gonads has been applied in such instances (because of their macroscopic appearance). If these gonads are intra-abdominal, there is 15-30% risk of malignancy [1]occurring by the time the young woman reaches her mid 20's. Cancer changes can occur in

childhood. These streak gonads usually carry minimal hormone producing capacity and do not contain any eggs or sperm. (In contrast normal ovaries produce hormones and contain thousands of follicles/ eggs providing fertility potential).

2. Complete Androgen Insensitivity Syndrome (CAIS). All people with this condition have XY chromosomes, but look and almost always identify as women. They do not have a uterus or ovaries. They have testes that produce testosterone (male sex hormone) in large amounts, but their body cannot respond to this because their androgen receptors (which are required for testosterone to act on a tissue) do not function. A consequence of this is that their testes are not fertile.

In the past, these testes were removed because the risk of cancer was believed to be higher than it is now known to be (2%)[1]. Today, girls and young women with CAIS would have their testes left in place, as the testosterone they produce can naturally be converted to oestrogen in the body. This allows them to develop breasts without the need for hormone replacement. When the young woman is in her late teens or adulthood, discussion may occur re potential removal of the testes– either due to the testes causing pain with sexual intercourse, or if the woman's preference is to definitively remove the increased cancer risk.

3. Partial androgen insensitivity syndrome (PAIS). People have XY chromosomes and testes that produce testosterone but their body is only partly responsive to this hormone. There is a considerable spectrum – with some people being born with almost normal male external genitalia, and others having almost normal female genitalia (but all will have no uterus). If the testes are undescended and inside the abdomen, the cancer risk of the testes is reported to be 50% [1]. These testes, although producing testosterone, are not capable of producing sperm. It is unclear whether in future there may be the capacity of using assisted reproductive technologies to extract sperm precursors - but current medical research, knowledge and understanding cannot offer any fertility potential to someone with this DSD.

Fertility is an issue for a number of other individuals with DSDs where gonadal function is not the problem, but rather their other anatomical problems. In some of the conditions including bladder exstrophy, cloacal exstrophy or cloacal anomalies, the significant structural anomalies may make pregnancy too challenging. In some of these conditions and other DSD conditions where females are born without a uterus, the only possibility for fertility is with the use of a surrogate to carry their pregnancy. Likewise for males with some of these penile structural conditions, penetrative sexual intercourse may be challenging (although assisted fertility techniques can be used to achieve fertility).

Gonadectomy

Current practice is that gonads are not removed in someone with a DSD if the gonads have any fertility potentially – thus these procedures are not sterilising procedures.

Today, very careful consideration is given prior to considering removal of gonads in someone with a DSD. The key reason for gonadectomy being performed relates to their malignancy risk. This is particularly the case for gonads that are neither hormone producing nor providing the prospect of any fertility. In such a scenario, if a decision for removal of a gonad or gonads is made, this only occurs after wide discussion and consultation. At a professional level, at RCH this discussion involves both the multidisciplinary

DSD team meeting, followed by discussion at the clinical ethics response group. Finally legal opinion is sought as some cases require authority from the Family Law Court of Australia under current caselaw.

Where possible, discussions concerning potential gonadectomy involves the young person with a DSD and seeks their input. Nonetheless, as the risk for malignancy in some of these conditions can reach 30% or even 50%, some of these decisions are made at an age when a young person is not old enough to fully understand the implications of surgery or have input into this decision. The decision in these cases clearly involves the parents acting on behalf of their child.

In some young women with specific DSDs that relate to problems with testosterone production, an adolescent girl or young woman would have looked like a girl at birth, but either due to lack of periods as an adolescent (due to an absent uterus), or due to development of excess hair in early puberty she is then found to have XY chromosomes. In this person there has not been enough active testosterone produced during fetal development to result in a male appearance of the genitals. At puberty the higher levels of circulating hormones may mean they begin to get extra hair, growth of their clitoris and/or possible get deepening of their voice. In addition, some of these conditions have a substantial risk of malignancy (28%-50% risk in 17BetaHSD)[1, 2]. In the past, a decision regarding gonadectomy may have been made reasonably rapidly. This was done to remove the testes (and thus the source of hormones which were considered counter to the individual's sex and gender of rearing), and which also had the increased malignancy risk. Today the pathway is far more careful as it is recognized that some individuals with a DSD may want to change their gender identity or wish to identify as indeterminate or intersex. In this scenario, although these gonads are not likely to carry any fertility potential, their hormone production may be beneficial and so the young person may choose to retain them, to facilitate a change in desired gender identity. This decision-making process takes time and thus any decision regarding possible gonadectomy would not be made until an informed and considered decision can be made by the person themselves.

The potential difficulty with this more conservative approach is that for some young people (e.g. those who definitively identify as female and do not wish to retain their testes), the perceived delay in surgery and the associated need for gonadal surveillance (with ultrasound or MRI) can be very frustrating.

The decision as to whether or not to remove gonads that are hormone producing, but not fertile, is now only undertaken after lengthy, sensitive and careful discussion. Whenever possible, the affected person themselves is involved and empowered to drive that process.

Sexual function

Sexuality is an important aspect of health and wellbeing. For positive sexual relationships, general quality of life, positive self esteem and body image are all contributing influences. People with chronic medical conditions who require repeated hospital attendance and daily medications, or who have had significant surgery, are at risk of poorer self esteem, and body image and reduced quality of life [3]. For many people with DSDs, repeated hospital attendance for monitoring of hormone levels, growth and development as well as surveillance monitoring of gonads with malignant potential is routinely required. Additionally some people with DSDs may have had genital and/or other surgery.

The term congenital adrenal hyperplasia (CAH) describes a number of conditions that result in abnormalities of hormone production in the adrenal glands. Due to high levels of androgen exposure in utero, women born with the most common form of CAH (21 hydroxylase deficiency) usually have some degree of virilization of their genital appearance – from subtle changes through to a male like external genitalia (despite the presence of normal uterus and ovaries). Reconstructive surgery to reverse these changes is frequently undertaken. The indications for this surgery depend on the degree of virilisation, but may include correction of urinary flow, opening of the lower vagina and/or clitoral reduction to establish anatomy that is congruous with female sex of rearing. International guidelines define the degree of virilisation for which surgery may be indicated; however the optimal timing of this surgery is not known [4] . This is because there are no data to support one particular approach (e.g. surgery in early infancy) over another (e.g. surgery in late infancy, mid-childhood, early adolescence or late adolescence). At RCH, surgery is most commonly performed in infancy. As part of our DSD multidisciplinary team we are fortunate to have highly skilled paediatric urologists with extensive experience in this area. We acknowledge that controversy exists around the optimal timing of surgery. The combination of evidence of satisfactory/good anatomical and cosmetic outcomes in our patient population who have had early surgery[5], the evidence of potential negative impacts of delaying surgery in terms of psychosexual wellbeing[6, 7], plus the clinical experience that surgery undertaken in teenagers is actually more difficult, all impact on this decision and the continuation of this surgical practice for those individuals where it is indicated. Parents are informed that differing opinions exist and are aware that deferring surgery is an option they can chose. All cases are discussed by the multidisciplinary team and the clinical ethics group prior to surgery.

Thus reports of reduced sexual enjoyment in people with DSDs may relate to a range of factors.

- The impact of their underlying condition. For example, some men with DSDs that result in reduced virilisation of the external genitalia have difficulties with penetration, while women with CAH who required surgery may have discomfort with penetrative sex.
- Sensory outcomes in women with DSDs, comparing those who have not had surgery with control women show poorer outcomes in those with a DSD, suggesting that some of the reduced sexual function relates to the condition or to aspects of care, rather than surgery[8].
- Reduced or altered sensation following previous genital surgery may relate to the exact surgical technique used, as this varies around the world[9]. Clitorodectomies have not been performed in Melbourne for over 40years but reports exist that suggest it was still performed in parts of the UK and the USA even in the 1980's[10, 11]. The technique used at RCH involves preservation of the neurovascular bundle. Internationally the experience of surgeons undertaking this procedure varies with some surgeons performing the procedure infrequently[8]. Likewise, the reported cosmetic and anatomical outcomes following genital surgery vary widely[5, 10, 12].
- Timing of surgery may have a negative impact. Although there is no direct evidence regarding the timing of genital surgery in girls, there is evidence from studies on boys. These report better self esteem and body image, and more positive attitudes towards intimate relationships in adolescents and young men if their genital surgery is completed before the age of 3years, compared to surgery in mid-childhood. Although some people advocate leaving all genital surgery till later when the person can consent themselves to the procedure, there are no studies to demonstrate a comparison of outcomes with this greater delay[6, 7].
- Lack of opportunity to discuss and explore sexuality and understand genital variation.

- Lack of access to clinicians in the adult setting who are familiar with the genital variations and potential problems in young adults with a DSD
- In some DSDs structural anomalies may directly impact on sexual function. For women born without a vagina and for those with bladder exstrophy, cloacal exstrophy or cloacal anomalies, a vagina needs to be made with dilators or with surgery. There are significant challenges in achieving a neovagina with no internationally accepted best technique.

Many of the changes that have occurred in the approach to caring for someone with a DSDs (as outlined above) have resulted from increased recognition of the difficulties that affected individuals have experienced and reported. Our multidisciplinary approach aims to enable sensitive exploration of body, self confidence, self esteem, and to optimise the opportunities for intimate relationships for an individual with a DSD. Nonetheless, follow-up studies to assess the impact of our current approaches on outcomes in various subgroups of DSDs are required.

Conclusion

The care of someone with a DSD is complex and requires a multidisciplinary team of health professional to support and care for the individual and their family. This approach has been in place at RCH Melbourne for a number of years, although as a specialized medical service it is not specifically funded.

Changes in medical and surgical knowledge combined with the recognition of the importance of adequate psychological support and the need for sexual health issues to be incorporated into care means that services provided today are different to those provided in the past.

We do not consider that forced or involuntary sterilization relates specifically to youth with DSDs, as it is rare that gonads removed for an increased risk of malignancy in this cohort have fertility potential. Where fertility is possible, every effort is made to preserve this potential.

Changes established in recent decades aim to optimize outcomes for affected individuals; however we acknowledge that outcomes related to current approaches remain to be established. In addition, areas of contention exist as to best practice in some aspects of DSD care. Funding for further research is required to better define optimal management strategies in certain DSDs.

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