Patient-Centered Care for Children with Disorders of Sex Development: A Case of 46,XY DSD in a 3 year old

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Disorders of sex development (DSD) refer to an extensive range of conditions wherein the chromosomal, gonadal or phenotypic sex is incongruous. Presented is a case of ambiguous genitalia in a 3 year old child reared as female since birth and subsequently assigned a male gender after being diagnosed with 46,XY DSD. Management ideally involves a multidisciplinary approach to address the myriad of issues in the care of a child with DSD and to address significant concerns including: gender assignment and gender identity; timing of medical and surgical interventions and equally important, psychosocial and psychosexual development. Current recommendations revolve around patient-centered care which advocates respect for the autonomy and personhood of the patient by postponing the making of crucial decisions, whenever possible, until the child is capable of participating in the decision-making process.

Key words: disorders of sex development, ambiguous genitalia, gender identity, gender assignment

Introduction

The birth of a healthy child after nine months of waiting is considered a fitting conclusion to a long process and deemed most rewarding. The anticipation among would-be parents builds up from the time of confirmation of a pregnancy to even beyond the aspirations set for the future of the child. As early as the prenatal stages, the initial question is whether the forthcoming child is a son or a daughter. While modern diagnostics can provide a good peek of the baby's sex, these procedures are not free from errors. Confirmation takes place only after the delivery of the child with the appreciation of the external genitalia as being that of a male or a female. In some instances however, the birth attendant cannot always give a straightforward response to the question. Such is the case in children born with ambiguous genitalia. Confusion, then concern often follows for the sake of the child's well-being. Since in a setting wherein being different may lead to segregation, a tendency for isolation or even ridicule is often dreaded.

Concerns on rearing, surgical, medical and psychosocial well-being of the child with DSD can be overwhelming. Rightly so, this should be addressed by medical health professionals in the best possible way. Needless to say, a medical situation with multiple issues of concern such as DSD is best dealt with by a team of professionals prepared and trained to address all aspects of the condition.

Case Protocol

The three year old girl RP was referred to the Reproductive Endocrinology and Infertility Clinic for ambiguous genitalia. She has had an unremarkable medical history so far. The patient was born to a then 30 year old housewife with uncomplicated prenatal history via spontaneous vaginal delivery in a local hospital. Prenatally, the mother was only seen twice at a local health center with no baseline prenatal ultrasound done. The mother denied exposure to radiation, contracting diseases during pregnancy, or intake of medications other than iron and multivitamin supplements. The patient's ambiguous genitalia was noted at birth. At the time, the labiascrotal structures had lesser volume and smoother appearance while the clitoro-penile structure was much smaller. Unfortunately, no investigations were done due to financial constraints. Newborn screening was advised but the parents

failed to comply. She was given a female name and registered as such in her certificate of birth. There were no significant medical developments during infancy. During the 3 years of life, the patient was reared as a female.

She is the youngest of three children with apparently normal female and male siblings, sired by a then 44 year old healthy market vendor. Both parents denied neither similar conditions nor any serious diseases in the family. Her motor, cognitive, behavioural and social developments were at par for her age. The patient was virtually healthy with no serious complaints. The parents, however, observed her preference to urinate in the standing position.

On initial consult, the patient was dressed in girls' clothes and was made up like one. Her vital signs were stable. She stood 99 cm and weighed 13.2 kg, both within the normal standards for pediatric Filipinos her age. Systemic physical examination revealed prepubertal (Tanner stage 1) breast development and pubic hair distribution. She did not exhibit neck-webbing and there were no areas of significant hyperpigmentation on the skin. No palpable abdominal masses were noted. On evaluation of the genitalia, palpation revealed a 2cm x 2cm doughy mass within the right labioscrotal fold and a smaller 1cm x 1cm doughy mass within the superior aspect of the left labioscrotal fold near the inguinal canal. Both labioscrotal folds exhibited ruggae and were more pigmented compared to the surrounding skin, with the texture and pigmentation more defined on the right. There was a phallic structure measuring 2.2 cm with a single ventral opening proximal to the perineum (Prader type 3). Digital rectal examination was deferred (Figures 1-3).

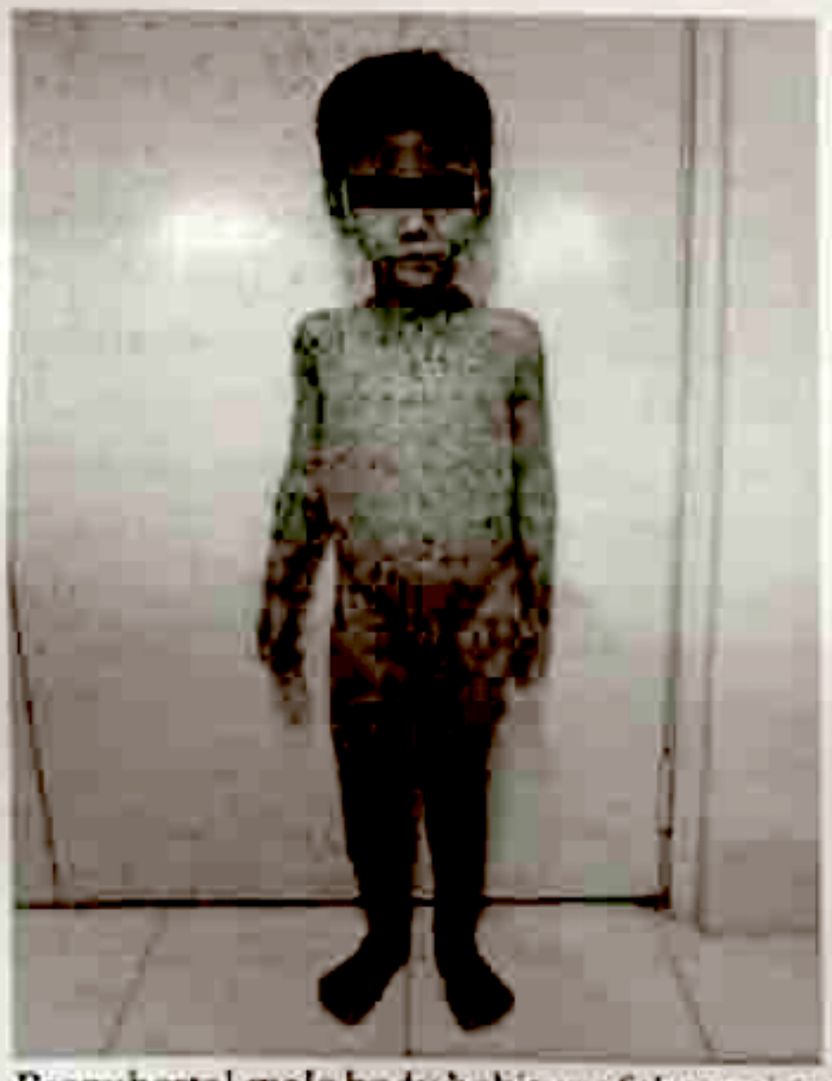

Figure 1. Prepubertal male body habitus of the patient on follow up after shifting gender assignment from female to male.

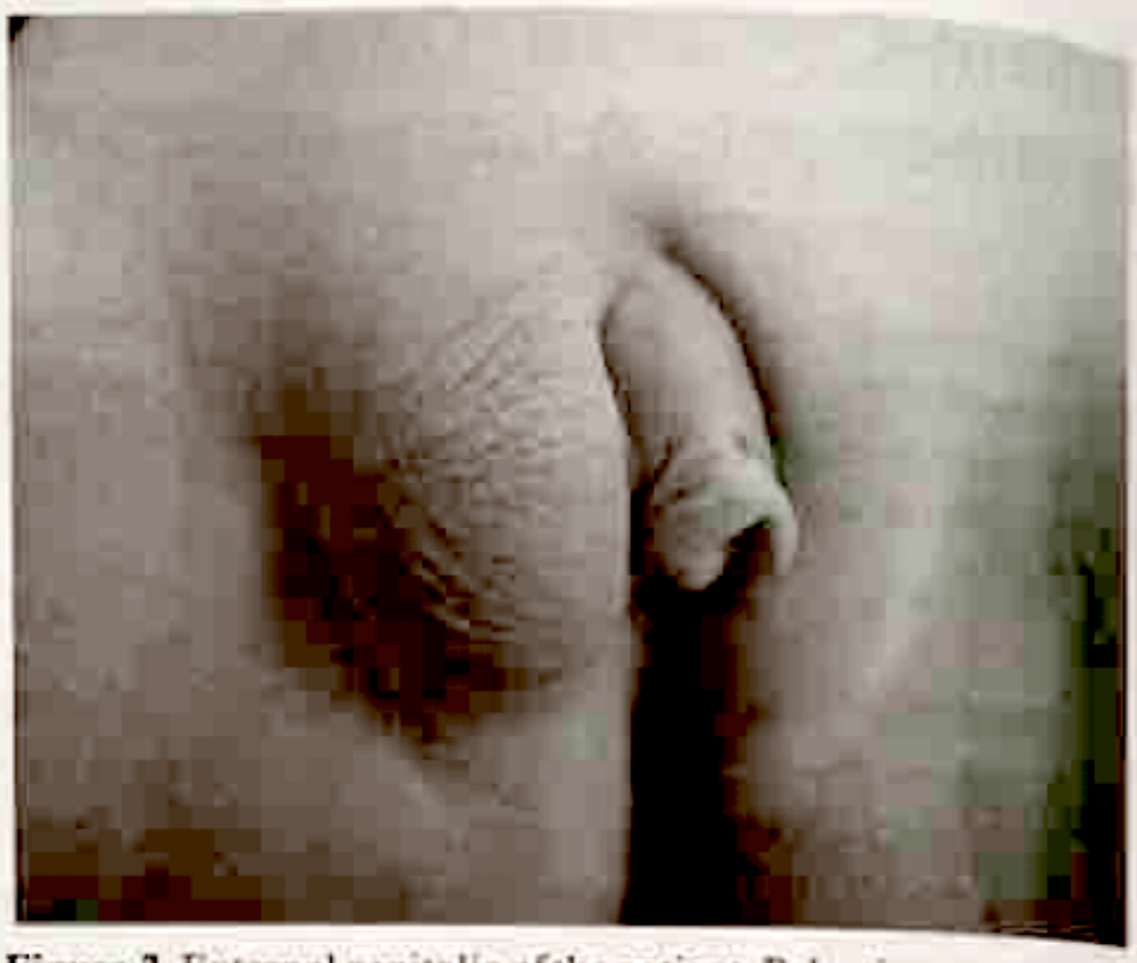

Figure 2. External genitalia of the patient. Palpation revealed a 2cm x 2cm doughy mass within the right labioscrotal fold and a smaller 1cm x 1cm doughy mass within the superior aspect of the left labioscrotal fold near the inguinal canal. The ruggae and hyperpigmentation were more prominent on the right labioscrotal fold. Note the phallic structure measuring 2.2 cm.

Figure 3. A single ventral opening (hypospadia) proximal to the perineum.

Initial assessment was Disorder of Sex Development (DSD). Further work-up was requested. Transabdominal and transperineal ultrasonography revealed the absence of a uterus and ovaries. There was a cystic area in the right labia within which was a 2.1cm x 1.3cm homogenous ovoid mass. Investigation of the contralateral labia revealed another cystic structure within which is a 2.5cm x 1.0cm central ovoid echogenic structure with a pedicle. No intraabdominal masses were noted and both kidneys appeared normal. These findings were signed out as suggestive of extrapelvic gonads (Figures 4-6).

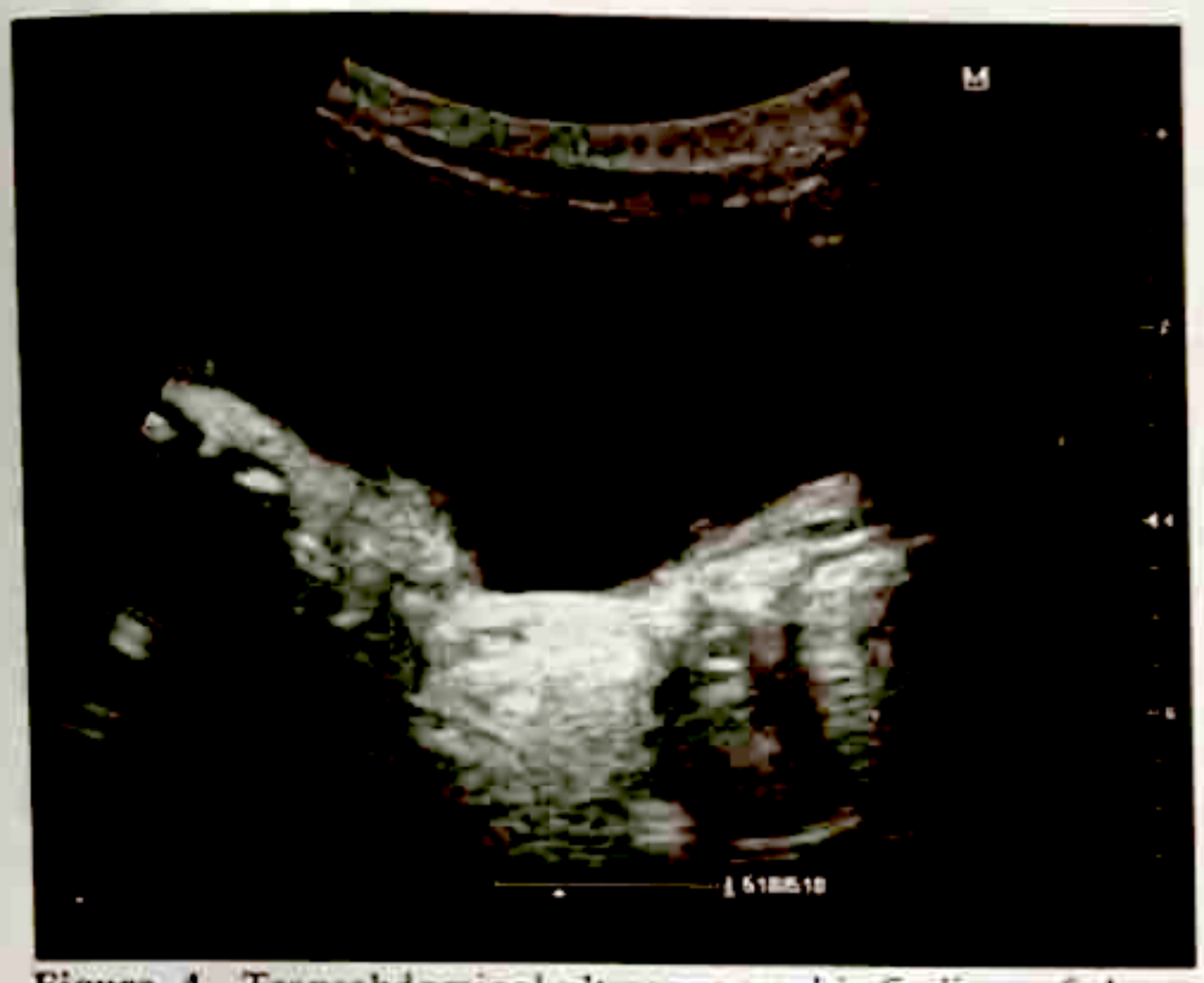

Figure 4. Transabdominal ultrasonographic findings of absent uterus and ovaries.

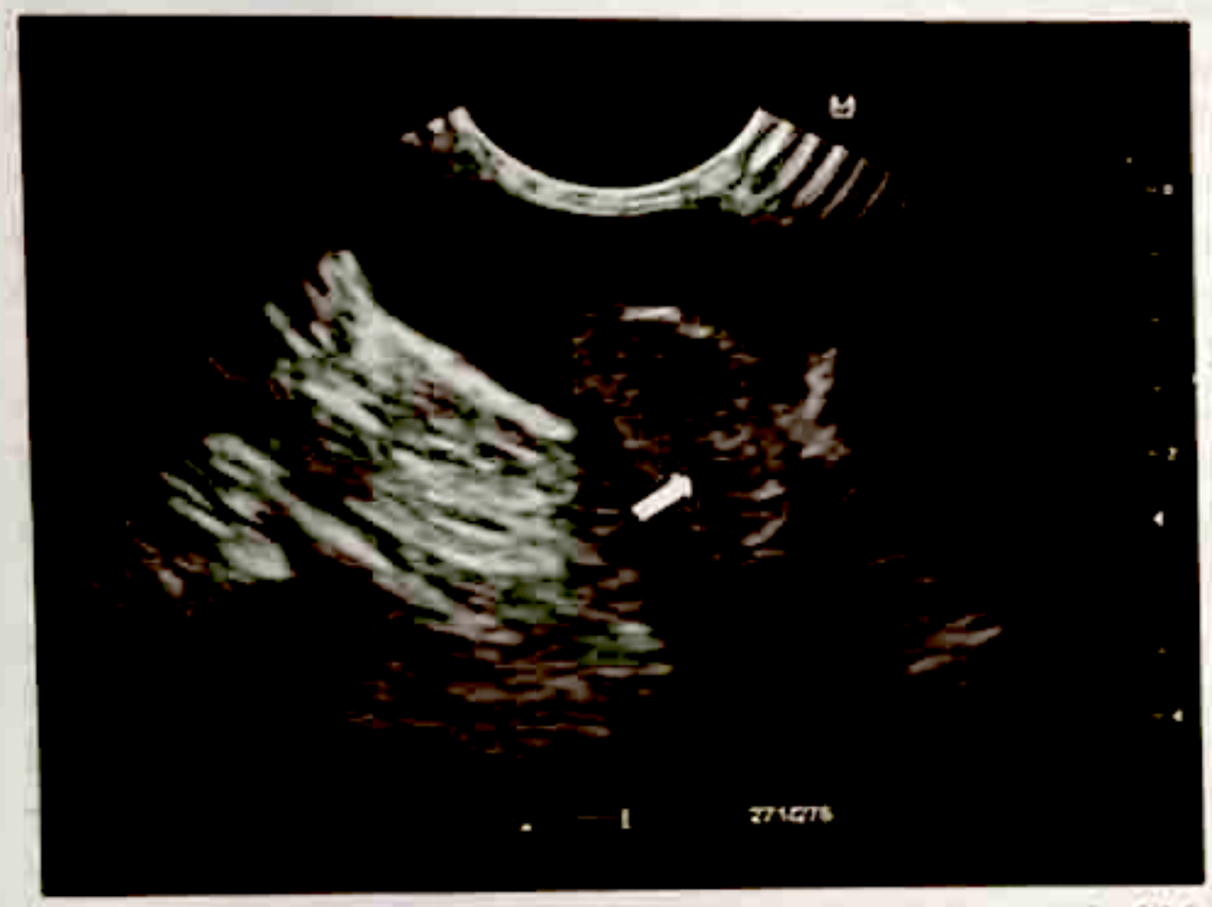

Figure 5. Transperineal ultrasound showing the right gonad within the labioscrotal fold.

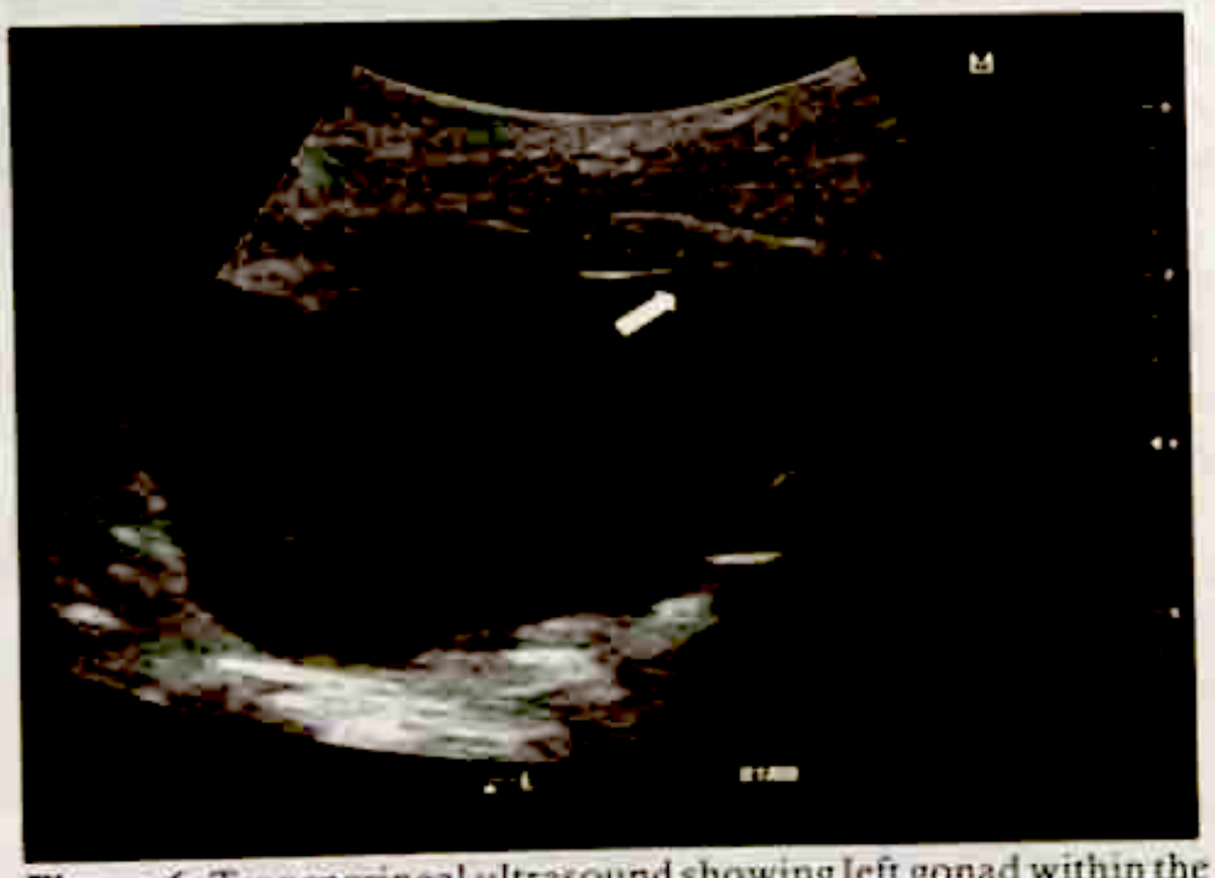

Figure 6. Transperineal ultrasound showing left gonad within the left labioscrotal fold near the inguinal canal.

Genetic studies revealed normal male karyotype (46,XY). Baseline hormonal studies showed testosterone of 0.40 nmol/L, FSH of 3.8 mIU/mL, LH of 0.08 mIU/mL and estradiol of 22.7 pg/mL. With these ancillary procedures, final diagnosis was 46, XY DSD.

The results of the diagnostics were disclosed to the family and the patient's genetic sex was confirmed. They were advised on the need for a multidisciplinary approach in management. Referrals to Pediatric Endocrinology, Child Psychiatry, Pediatric Surgery and Urology were made. The patient however failed to follow up and has yet to be seen by the other services. When the family was contacted to bring the patient back to the clinic, she already had the physical appearance of male child (cropped hair, dressed up in boys' clothes) and was already given a male name.

The parents raised their concerns regarding rearing of the patient and the confusion surrounding the matter. A general supportive advice given was to rear the patient based on the typical gender behaviour and characteristics being exhibited. That is if her, now his, behaviour was consistent with that of a male child, then the patient may be reared male and vice versa until the time the patient is able to decide for himself which gender he would later preferred to be assigned. This advice, however, proved to be difficult for the family to follow.

Discussion

Ambiguous Genitalia

Up until the 9th week of gestation, the fetus remains in a bipotential state wherein the neutral primordia of the external genitalia is influenced by steroid signals produced by the gonads to develop either male or female genital structures. Masculinization of the external genitalia is the end result of the testosterone coming from the Leydig cells in the testicles. This results in the formation of the typical male external genital composed of the penis, penile urethra and the scrotum. Conversely, absence of strong androgen influence in females result in the formation of the typical female external genitalia composed of the clitoris, major and minor labia, and the lower vagina and urethra.[1]

Ambiguous genitalia in newborn infants are a social as well as a medical emergency. There is the urgency in deciding the appropriate sex of rearing and the necessity of preventing potential life-threatening metabolic disturbances that might be associated with some of the underlying causes for the condition. Conditions that may result in death thus requiring immediate intervention (e.g. salt-wasting type of congenital adrenal hyperplasia).[2]

Disorders of Sex Development

Disorders of sex development (DSD) refer to an extensive range of conditions wherein the chromosomal, gonadal or phenotypic (anatomical) sex is discordant. It is reported to occur in 1 of every 4,500 births.[3] DSDs are defined as conditions involving the following elements: congenital development of ambiguous genitalia, congenital disjunction of internal and external sex anatomy, incomplete development of sex anatomy, sex chromosome anomalies and disorders of gonadal development.[4]

In 2006, a consensus statement on the management of intersex disorders was proposed by the Lawson Wilkins Pediatric Endocrine Society and the European Society for Pediatric Endocrinology to classify disorders of sexual development. This modified nomenclature is the product of progressive understanding of the molecular basis of atypical sex, the enhanced ethical issue awareness, as well as patient advocacy concerns.

Sex chromosome DSD are associated with abnormal karyotyping which leads to abnormal gonadal development. This includes 45, X (Turner and variants), 47,XXY (Klinefelter and variants) and 45X/46XY (mixed gonodal dysgenesis), choromosomal ovotesticular DSD (46XX/46XY chimeric type or mosaic type).

The 46, XY DSD previously called male pseudohermaphroditism is secondary to disorders of testicular development (complete gonadal dysgenesis, partial gonadal dysgenesis, gonadal regression, ovotesticular DSD) and androgen synthesis or action deficiency (androgen synthesis defect, LH-Receptor defect, androgen insentivity syndrome, 5α-reductase deficiency disorders, AMH timing defect, endocrine disrupters, cloacal exstrophy).

Conditions previously termed female pseudohermaphroditism falls under 46, XX DSD which result from abnormal gonadal development (ovotesticular DSD, testicular DSD, gonadal dysgenesis) or androgen excess from fetal (congenital adrenal hyperplasia), maternal (gestational hyperandrogenism), or fetoplacental origin (aromatase deficiency, P450 oxidoreductase deficiency).[1,3,5]

Clinical Evaluation

Thorough clinical history taking is the first step in the evaluation of a patient with DSD. Relevant prenatal history should be investigated including maternal exposure to androgens or medications acting as endocrine disruptors, and signs of maternal gestational virilization. Family history of similar conditions like sudden unexplained infant demise or conception between consanguineous couples should be investigated.[1] In the case presented, there was no history intake of such medications nor report of similar conditions in the family of either parent.

A detailed physical examination and documentation of the genitalia is necessary to evaluate the degree of genital anomaly. Focus is given on the examination of the gonads, phallus, chordee, epispadias and hypospadias, labioscrotal fold texture and pigmentation, and anogenital distance.[6] The patient in this case presented with bilateral labioscrotal folds exhibiting ruggae and hyperpigmentation within which are masses assumed to be the gonads. A phallic structure with perineoscrotal hypospadia was also noted.

Essential in the evaluation of ambiguous genitalia is karyotyping to determine the chromosomal sex. Hormonal studies to measure the adrenal and gonadal steroid secretion may include 17-hydroxyprogesterone (baseline and ACTH stimulated) which expedites exclusion of the life-threatening 21-hydroxylase variety of congenital adrenal hyperplasia, 11-deoxycortisol and DHEA to identify less common forms of CAH, testosterone (baseline and human chorionic gonadotropin stimulated) and dihydro-testosterone which are useful in diagnosing chromosomal male DSD and basal measurements of LH, FSH, AMH to assess testicular function.[1,2,3,7]

Critical in the investigation of a child with ambiguous genitalia and essential in depicting the internal organs and urogenital anatomy is imaging. The primary modality in establishing the presence or absence of gonads and derivatives of the mullerian system is ultrasonography and should include the inguinal, perineal and adrenal regions. In cases of 46, XY DSD, the testes may be found within the inguinal canal or the abdominal cavity. Other findings may include absent uterus and ovaries in the pelvis. In other forms of DSD, genitography may be used to demonstrate the urethra, vagina or any fistulous or complex tracts while magnetic resonance imaging may be reserved for more difficult cases.[7]

Our patient has a normal 46, XY karyotype with bilateral palpable testes limiting our diagnosis to chromosomal male DSD. The patient's testosterone is consistent with prepubertal levels. Ideally, an hCG stimulation test should be performed where a satisfactory response in penile length will be highly suggestive of partial androgen insensitivity and an elevated testosterone to dihydrotestosterone ratio will point to the diagnosis of 5-α-reductase deficiency. Sonologic findings are consistent with the clinical suspicion of the palpable gonads and confirmed the absence of uterus. Considered concurrently, we are able to arrive at the diagnosis of 46,XY DSD (disorder of androgen sensitivity or action). Further studies should provide further enlightenment on the more specific etiologic diagnosis.

Management

The objectives of management of complex genital anomalies should address issues that incorporate the establishment of a stable gender identity, psychologically well-adjusted patient and family, education on the condition that is appropriate for the age of the patient, optimal sexual function, optimal body image, maximized fertility potential and interventions employing the least number of medical and surgical intervention that will allow the patient and family to achieve the highest quality of life.[5]

There is a need to identify the underlying cause of DSD to provide a tailored approach to management. Confirmation of the etiology may help with the treatment of related hormonal deficiencies and assist in the anticipation of the immediate and long term medical concerns. It is necessary for parents and relatives of patients with DSD to understand what brought about the condition allowing them to plan ahead.[6]

In 2006, the Intersex Society of North America (ISNA) proposed clinical guidelines which represent an ideal management of DSD in childhood. Emphasis was placed on patient-centered care which focuses on the well-being of individual patients. They proposed a multidisciplinary approach to address the multidimensional issues involved in DSDs for provision of diagnosis, treatment and support to the patients and their family. This multidisciplinary team preferably consists of members from child psychology/psychiatry, genetics and genetic counselling, gynecology, nursing, pediatric endocrinology, pediatric urology, social work and others as needed.

The protocol begins with the obstetrician-gynecologist or attending health care professional who first identifies DSD in the newborn. Initial findings should be relayed to the family and reassurance offered. It must be explained that further consultation with professionals familiar with the condition is necessary. A team liason officer then meets with the family and the family pediatrician while initial supportive counselling is being provided by a trained health professional. The team coordinator contacts the members of the multidisciplinary team for a case conference after which a small number of representatives proceed with examination of the child with the family and family pediatrician involved in every step of the process. Genetic, endocrinologic and imaging studies are ordered to ascertain the etiology and relevant details of the child's anatomy and physiology which will aid in the initial gender assignment and planning for long-term care.

A multidisciplinary case conference is then called to discuss the clinical findings and available laboratory results to come up with initial gender assignment recommendations considering the gender the child will likely identify with when older. Treatment options as well as short- and long-term follow up plans are proposed then discussed with the family allowing sufficient time for them to come up with the decision. During the entire process, psychosocial support attended by trained health care professionals is offered to the family.

In cases of DSD diagnosed after the newborn period, decision making is less urgent if the child is medically stable. This allows sufficient time for the team to order diagnostics, hold case conferences, and formulate recommendations to the family. While in the process of awaiting the diagnosis and the recommended plan of management, psychosocial support is continually provided to assist the anxious family. During physical examination, the young child should be aware of what is transpiring, explained in age-appropriate terms and performed by the least number of team members as possible to prevent the feeling of being overwhelmed. In the more mature child, peer support including family-to-family support should be offered. The team should search for evidence of patient and family being socially- or emotionally-at-risk and provide prompt intervention as necessary. Furthermore, in the event that the initial gender assignment is not congruent to the child's gender identity, the team should provide appropriate support for the child and the family during social gender transition.

Gender Identity and Gender Assignment

A person's gender identity is one's concept of self as being a female or a male. It is determined by the chromosomal, gonadal and phenotypic sex. That is, a person develops an identity of being masculine or feminine due to the sum of the influences of the sex chromosome (typically XX or XY), the direction to which the gonads differentiate (ovary or testes), and whether the external genitalia appears morphologically female or male. An equally important determinant of gender identity is the influence of the environment (social and cultural factors) the individual belongs.[1]

One of the most confusing matters in DSD is with regards to how the affected child should be reared. Gender assignment in the newborn with major congenital anomalies has serious implications on the succeeding medical and surgical interventions and equally important are the consequences on the patient's psychosocial and psychosexual development.

Assigning the sex of rearing is the parents' right, obligation and responsibility and they should be encouraged to maintain a perspective that sexual features and gender is a continuum. While the hope is for the gender identity to be consistent with the initial gender assignment,

the child's development of the former which is incongruent to the initial assignment cannot be regarded as a failure.[9] The considerations for gender assignment include specific details of the clinical syndrome, genital anatomy and surgical options, potential for fertility, potential for adult sexual response and factors that may predict the most likely adult gender identity. The decision should be based also on the best prognosis for psychosocial and psychosexual function and health of the patient. Furthermore, cultural factors, education and society play a major role in the outcome of such assignment and should be attributed considerable weight.[8,9]

On one hand, the predominant role of gender assignment in gender identity formation has been shown by some studies in patients particularly in cases of partial androgen insensitivity (reared either as male or as female and subsequently developed gender identity concordant with assigned gender) even in the absence of strong hormonal influence.[10] On the other hand, the effect of prenatal androgen exposure has been established in studies involving genetic males exhibiting high likelihood of recognizing male sexual identity apparently independent of sex-of-rearing. Gender identity in this set of patients is not dependent of the morphology of the genitalia. Unpredictable sexual identity outcomes, however, were noted in genetic males with inadequate prenatal androgen effects or genetic females experiencing inappropriate androgen effects (from CAH).[11]

Interestingly, genital appearance may not be as crucial a determinant in gender identity as once thought. Based on long term outcome of case studies involving males with complex genital anomalies, male gender identity does develop in the absence of gender specific genitalia.[8] Our index patient is presently being reared as male which is not entirely wrong. While recent evidence supports rearing the chromosomal male with DSD as male, the family should remain flexible and open to the developing gender identity of the child. That is, rigidity should be discouraged and the child be allowed to develop the identity that is most "natural" to him or her.

The 2006 guidelines by the ISNA advocate supporting the parents in realizing that each child is unique, acceptable and lovable. Obviously, it is unsound to assume that one can accurately determine the gender identity a child with DSD will grow up with as puberty is approached. Examinations and investigations are employed to shed light on the etiology and the prognosis of DSD with concerns in the anatomy and physiology. The valuable information acquired will assist the family in assigning the most appropriate gender to their child.

The family should be educated on what is known regarding gender identity outcomes in similar cases for them to come up with the best decision. It should be stressed, however, that in some cases, a child may manifest behaviour and develop an identity atypical to the assigned. If such is the case, psychological support and access to resources including support groups or talking with families sharing the same experience should be offered as these may aid in the adjustment the family may have to make.

When To Give Medical Therapy

Hormonal induction of puberty should attempt to assimilate normal maturation at the expected age of puberty to induce secondary sexual characteristics, pubertal growth spurt and optimal bone mineral accumulation.[6] Consistent with the general recommendation of the 2006 guidelines of the ISNA, medical or hormonal therapy is best delayed until the child develops the cognitive capacity to participate in the decision making. One reason for this is that the child has a developing gender identity and the effects of exogenous hormones may produce results incongruent with what the child identifies with. Administered hormones are not without potential risks and it is most appropriate if the patient understands the risks before making a decision.

When To Do Surgical Intervention

Previously, surgical interventions were performed as an adjunct to reinforce the initially assigned gender to the child. Parents and families of children with DSD approve surgery, with or without the "consent" of the young child, supported by the noble intention of sparing their children the shame and psychological distress that the condition entails. However, recent evidence and accounts from actual DSD patients showed that psychological trauma from the surgery itself was confounded by the feelings of betrayal upon discovering that surgery was performed without their consent. Additionally, a significant number of DSD patients who underwent early surgery to be congruent anatomically to the gender of assignment were distressed when the gender identity they developed was inconsistent with their created anatomy. For these reasons, it is best to allow the patient to take part in the decision on whether or not surgery should be done depending on the gender he or she identifies with. In many instances, the expertise of a child psychologist or psychiatrist may be necessary to assess the cognitive capacity of a child to partake in the decision-making. The patient and the family should be made aware of the documented risks related to the surgeries: reduced sexual sensation, sexual dysfunction, chronic pain and neoplasia in the neovagina. Once informed of such risks, when the patient and family decide to pursue surgery, the patient's autonomy should be

respected. While it is easier to reach a decision if the patient is cognitively capable of participating, the family of a younger patient may suffer anxiety in rearing a child with "ambiguous sex." Recent accounts and studies however have reported that the risk of psychosocial problems and maladjustments is no greater among such patients compared to the general population.

The general recommendation of the ISNA is to delay the surgery if there is no pressing threat until the patient can participate in the decision-making. Functional and healthy gonads should not be removed due to subsequent loss of fertility and benefits of endogenous hormones. There are, however, circumstances where immediate intervention is necessary - e.g. creation of a urinary outflow tract or removal of a malignancy. The risk of malignancy is highest in partial androgen insensitivity (~ 10 to 15% risk of germ cell tumors in testes not in the scrotum) and lowest in complete androgen insensitivity syndrome (~2%). If the estimated malignancy risk is low, surgery may be postponed until after the patient reaches puberty. Orchidopexy, in cases where in the testes are not within the scrotum, may be necessary to assess testicular development and malignancy risk by means of biopsy because the risk of malignancy increases as the testes is further away from the scrotum. If gonadectomy needs to be performed, testicular prosthesis must be discussed with age-appropriate patient. Surgical intervention should be discussed thoroughly between the family, the medical team, and most importantly the patient - if of appropriate age,.[6,12]

The good news is that current developments result in improved surgical outcome allowing reconstruction of the genitalia while maximizing preservation of neurovascular supply and erectile mechanism. The 2006 consensus statement by the Lawson Wilkins Pediatric Endocrine Society and the European Society for Pediatric Endocrinology recommends deferral of surgery for modest clitoral hypertrophy, early vaginoplasty and clitoroplasty in children with a high proximal junction between the vagina and urethra, and consideration of gonadectomy in those with risk of malignancy as previously mentioned.[9]

Psychosexual Concerns

A child afflicted with the condition may suffer from serious physical and psychological distress inherent to DSD itself. Moreover, the series of physical examinations, diagnostic procedures, surgery, medical therapy and long-term follow up complicate the suffering. Studies showed that these patients reported high degree of mental distress, high frequency of suicidal thoughts and impaired quality of life. Psychological counselling should be helpful in achieving the goal of development of gender identity consistent with sex of rearing. It should be available to all patients with DSD with the aim of improving quality of life and reducing the social and psychological problems.[9, 13]

Disclosure of Truth

The truth about the condition should be discussed at a level that is understood by the child, with the family present under the guidance of a trained mental health professional. The ISNA recommends full disclosure of medical facts to the child typically at the latest by 16 years of age so as to avoid the psychologically traumatic revelation when the child reaches the legal age of 18 - when he or she may have full access of the medical records. Full disclosure fosters trust and openness between the patient, the family and the doctor to facilitate the long-term management required by the condition. Allowing knowledge of the truth is a step towards patient empowerment. The more the patient and the family understand the condition, the better will be their adjustment and ability to live a better quality of life.

Conclusion

Presented is the case of a 3 year old with 46,XY DSD initially reared as female then shifted to a male sex assignment upon learning of the chromosomal sex. DSD is a challenging group of conditions involving medical and surgical dilemmas, and equally important psychosocial and psychosexual developmental concerns. Previous management practices are now being reconsidered as knowledge on molecular basis of diagnosis accumulates, ethical awareness increases and patient advocacy issues are considered. Recent recommendations focus on multidisciplinary approach to management to address the multidimensional issues inherent to the condition. While every treatment plan is individualized, the central advocacy is towards patient-centered care, as management decisions are delayed, whenever possible, until the patient is capable of participating in the decision-making process.

For RP and his parents, it was advised that his rearing be open and nurturing in character allowing him the opportunity to develop the gender he would grow to be most comfortable and satisfied with. Medical and surgical interventions, if eventually deemed necessary, must be timely to allow RP to acquire the necessary faculties to decide with confidence what is best for himself. For now, psychiatric support for both RP and his family is the most important intervention - not only for the meantime, but for his lifetime.

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