IJHRMLP
ISSN 2394–806X (Print), ISSN 2454-5139 (Online)

International Journal of Health Research and Medico-Legal Practice

CASE REPORT

Management of a child with disorder of sex development: social, ethical, and legal implications

Manuscript ID: *IJHRMLP-589*
Paul J[1], Dutta HK[2]
Address for correspondence:
[1]Postgraduate trainee
Department of General Surgery
[2]Professor (Corresponding author)
Department of Pediatric Surgery
Assam Medical College and Hospital
Dibrugarh, Assam
Mobile: +919435031257
Email: hemontdut@gmail.com
Article received: 01-12-2023.
Received (revised): 28-12-2023.
Editorial approval: 30-12-2023
Checked for plagiarism: Yes.
Peer-reviewed article: Yes.
Editor who approved:
Prof. NC Bhattacharyya

ABSTRACT

Disorders of sex development comprise a heterogeneous group of congenital conditions associated with atypical development of internal and external genitalia. There may be associated genetic variations and hormonal derangement. The affected children may present at birth with genital ambiguity or may present in later childhood or during adolescence. Disorders of sex development can be classified into several categories, including chromosomal, gonadal, and anatomic abnormalities. Controversy exists regarding the management of such children with social, ethical and legal issues that often influence the time and type of surgical procedures. Most importantly, patient management needs to be individualized, particularly for decisions related to sex of rearing, surgical interventions, hormone treatment, and potential for fertility preservation.

Keywords: *Ambiguous genitalia; intersex disorders; CAH; genitoplasty.*

Cite this article: *Paul J, Dutta HK. Management of a child with a disorder of sex development: social, ethical, and legal implications. Int J Health Res Medico Leg Prae. 2023 Jul-Dec;9(2):62-65. Doi: 10.31741/ijhrmlp.v9.i2.2023.9*

INTRODUCTION

A baby born with ambiguous genitalia becomes a cause of considerable apprehension and anxiety among the parents and family members. Assigning sex of rearing is a great responsibility for the medical team. However, this has significant social, ethical and legal implications. In such situations, the medical staff must respond with utmost care and sensitivity to the individual and the family. The term "Disorder of Sex Development" (DSD) has replaced the earlier "intersex disorders", "hermaphrodites", or "ambiguous genitalia" as it is a more inclusive terminology. We are presenting a child with female genotype and DSD, who had undergone feminizing genitoplasty and discussed in brief the potential social, ethical and legal issues.

CASE REPORT

A 4-year-old child reared as a female presented with DSD in the form of a phallus-like structure with posterior labial fusion. The child was first born, and the mother had an uneventful antenatal history except for hypertension. At three months of age, the child was diagnosed with asymptomatic hyperkalemia, and with further investigation, a diagnosis of congenital adrenal hyperplasia-classical variant (CAH) with raised 17-hydroxyprogesterone level due to 21- hydroxylase deficiency was made. Karyotyping revealed a normal female genotype (46XX). The child was put on daily oral hydrocortisone therapy and was asymptomatic following that. The child was referred for surgical opinion. On genital examination, a well-developed

phallus (4 cm) with proximal hypospadias was noted [**Figure 1 A and B**].

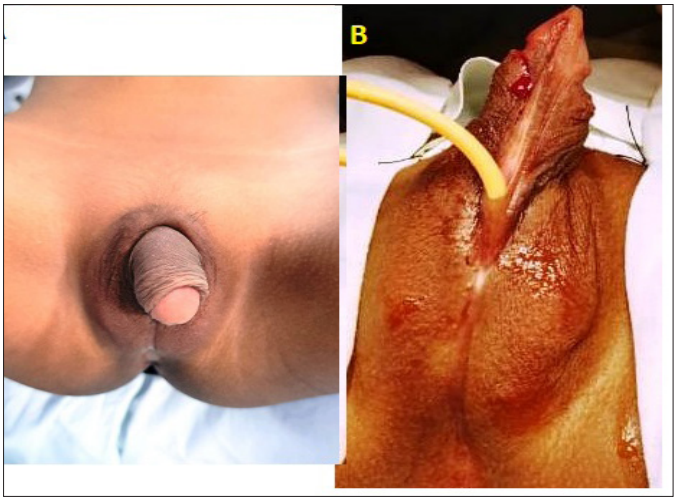

Figure 1 (A)Well-developed phallus and (B) with a hypospadiac opening

The scrotum was well formed but underdeveloped, and no gonads were palpable either in the scrotum or the inguinal canal. No other significant abnormalities were detected on systemic examination. Ultrasound of the abdomen was normal, with the presence of mullerian structures. MRI confirmed these findings. The parents and family members were counselled, and they agreed to the correction of genital abnormality. Surgery was planned for clitoroplasty, vaginoplasty and labioplasty. Penile skin was used for the construction of labia minora and vagina. Both corpora cavernosa were mobilized up to ischial rami and excised. The Glans with intact neurovascular pedicles were trimmed and fixed to the underlying tissue**[Figure 2**].

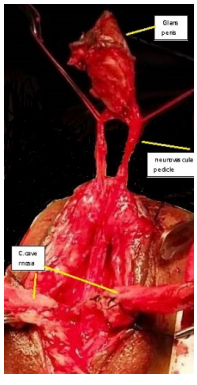

Figure 2 Dissected and mobilized neurovascular pedicle attached to the glans

The patient had an uneventful recovery and was discharged on day seven. She has been on regular follow-ups with cortisone therapy. The dilatation program of the neovagina with Hegar's dilator was continued for four months. The surgical outcome has been satisfactory [**Figure 3A and B**].

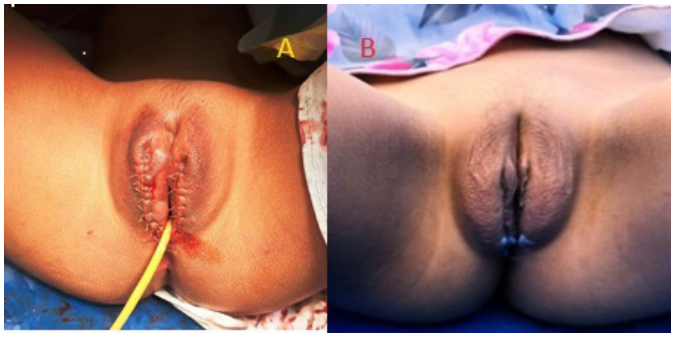

Figure 3 (A) After surgery and (B) after three months

DISCUSSION

A child's sexual identity can be expressed in various ways; genotypic sex deals with the genetic composition, phenotypic sex relates to the anatomy of the external genitalia, gonadal sex pertains to types of gonad present (testis, ovary, ovotestis, streak gonad, etc.). Studies have shown that DSD children do not always conform to the sex of rearing as they grow into adults.[1]Gender identity is the self-defined experience of one's gender. Gender identity disorder (GID), earlier known as gender dysphoria, refers to a conflict between a person's physical or assigned sex and the gender with which they identify.[2] Unlike DSD, a transgender person is born with unambiguous sexual anatomy, and their identity and gender do not correspond with their birth sex. Because of gender dysphoria, sex reassignment is often done during adulthood as per the individual choice. In contrast, a child with DSD is born with ambiguous genitalia and sex assignment surgery is done after birth, essentially for medical indication and after obtaining parental consent. In 2019, activists of transgender rights tried to include children with DSD as transgender; a lengthy legal battle ensued and in 2022, the Madras High Court directed the National Medical Commission to constitute a committee of experts to

IJHRMLP
ISSN 2394-806X (Print), ISSN 2454-5139 (Online)

formulate guidelines for the management of children with DSD to protect their rights.[3]

Meanwhile, Human Rights Watch, USA and other agencies raised issues of ethical considerations in the management of children with DSD, and they felt that intersex surgeries are unethical and should be avoided until the patient can actively participate in the decision-making process.[4]

Another critical aspect of gender assignment surgery in children with DSD is the cultural issues prevalent in society. Indian society is very sensitive to gender identity at birth, and any anomaly in a newborn baby's sexual anatomy may be a cause of immense anxiety and confusion among the family members. Studies have shown that DSD due to congenital adrenal hyperplasia, as in the present case, causes less gender identity disorder (GID), favouring the female sex of rearing in them.[5] In some other forms of DSD, sex assignment procedures are delayed till after adolescence depending on the child's anatomy, hormone profile and genetic composition.

Based on the national guidelines, surgery for DSD should be conducted at speciality centres and a local multidisciplinary committee (LMDC), including a paediatrician/endocrinologist, pediatric surgeon and psychologist/psychiatrist, should decide on modalities of treatment to be undertaken after a thorough evaluation and discussion with all stakeholders.[6] LMDC should keep a record of a signed document from all members and parents about the necessity for surgical intervention.

Feminizing genitoplasty is a complex and elective surgical procedure which includes clitoroplasty, vaginoplasty and labioplasty. Clitoroplasty is known to alter/impair sensation.[7,8] However, results are much better with the development of nerve-sparing techniques.[9] In the present case, the neurovascular pedicle supplying the glans was meticulously dissected and preserved. Some surgeons advocate delayed surgery when it can be safely performed with the informed consent of the adolescent girl.[10,11] However, many parents and caregivers feel satisfied with early surgery. Some of them want clitoromegaly due to CAH to be excluded from other intersex disorders. These should be considered separately in legislation about childhood genital surgery.[12,13]But controversy continues regarding the treatment of some severe forms of CAH when the child is reared as male, and late feminizing genitoplasty is considered unwise.[14]

In summary, surgical reconstruction for children with DSD should be individualized and considered after a thorough evaluation of the child. The decision should be taken by the local multidisciplinary team after discussing it with parents and family members. Long-term follow-up of these children is essential regarding their physical and psychosexual development.

DECLARATION

Certified that none of the authors have any conflict of interest in this work. The work is not funded by any agency.

REFERENCES

1. Yang JH, Baskin LS, DiSandro M. Gender identity in disorders of sex development: review article. Urology. 2010;75:153-9.
2. Cohen-Kettenis PT, Pfäfflin F. The DSM diagnostic criteria for gender identity disorder in adolescents and adults. Arch Sex Behav. 2010;39:499-513.
3. Justice Anand Venkatesh N. Sushma and Seema v. commissioner of police, 2021. p. W.P.No. 7284 of 2021 (Madras High Court).
4. Tamar-Mattis A, Ittelson A, Fraser S, Zieselman K. I want to be like nature made me: medically unnecessary surgeries on intersex children in the us. Human Rights Watch. (2017). [cited 2023 Dec1];

Available from: URL:https://www.hrw.org/report/2017/07/25/i-wantbe-nature-made-me/medically-unnecessary-surgeries-intersexchildren- us

5. Babu R, Shah U. Gender identity disorder (GID) in adolescents and adults with differences of sex development (DSD): a systematic review and meta-analysis. Pediatr Urol. 2021;17:39-47.
6. Babu R. Changing paradigms in intersex management: Legal, ethical, and medical implications. J Indian Assoc Pediatr Surg. 2022;27:372-5.
7. Abacı A, Çatlı G, Berberoğlu M. Gonadal malignancy risk and prophylactic gonadectomy in disorders of sexual development. J Pediatr Endocrinol Metab. 2015;28:1019-27.
8. Crouch NS, Liao LM, Woodhouse CR, Conway GS, Creighton SM. Sexual function and genital sensitivity following feminizing genitoplasty for congenital adrenal hyperplasia. J Urol. 2008;179:634-8.
9. Creighton S, Chernausek SD, Romao R, Ransley P, Salle JP.Timing and nature of reconstructive surgery for disorders of sex development – introduction. Pediatr Urol. 2012;8:602-10.
10. Ahmed SF, Achermann JC, Arlt W, Balen AH, Conway G, Edwards ZL, *et al.* UK guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development. Clin Endocrinol (Oxf). 2011;75:12-26.
11. Diamond M, Garland J. Evidence regarding cosmetic and medically unnecessary surgery on infants. Pediatr Urol. 2014;10:2-6.
12. Fisher RS, Espeleta HC, Baskin LS, Buchanan CL, Chan YM, Cheng EY, *et al*. Decisional regret about surgical and nonsurgical issues after genitoplasty among caregivers of female infants with CAH. J Pediatr Urol. 2022;18:27-33.
13. Szymanski KM, Rink RC, Whittam B, Hensel DJ, Braga LH, Donahue KL, *et al*. Majority of females with a life-long experience of CAH and parents do not consider females with CAH to be intersex. Pediatr Urol. 2021;17:210.e1-9.
14. Chowdhury TK, Laila K, Hutson JM, Banu T. Male gender identity in children with 46, XX DSD with congenital adrenal hyperplasia after delayed presentation in mid-childhood. Pediatr Surg. 2015;50:2060-2.