doi: 10.48095/ccachp20256

Experience with pediatric and adult cases of ambiguous genitalia reconstructed with a single stage feminizing genitoplasty procedure

N. Paul, S. Adhikari

Department of Plastic Surgery, Institute of Post Graduate Medical Education & Research, Kolkata, India

Summary

Background: The surgical procedure of feminizing genitoplasty aims at restoration of normal anatomy and function in various cases of disorders of sexual development with ambiguous genitalia. **Material and methods:** Between April 2021 and May 2023, 23 patients underwent a single stage feminizing genitoplasty procedure at the department of plastic and reconstructive surgery. All the patients underwent clitoroplasty with partial glans preservation, omega flap vaginoplasty and labioplasty. Cases with only clitoroplasty were excluded from the study. Of these 23 patients, 17 had congenital adrenal hyperplasia and the rest 6 had varying degrees of androgen insensitivity syndrome. The age of the patients ranged from 4 to 23 years and all were raised as females. The mean operating time was around 120 to 150 minutes and average hospitalization period was 7 to 8 days. At follow-up evaluation, no major complications were observed. In all cases the vaginal introitus was located in the physiological position and was of varying size and elastic. **Conclusion:** This procedure of single stage feminizing genitoplasty enables reconstruction with good cosmetic and functional results not only in children but also in adults presenting with ambiguous genitalia.

Keywords

congenital adrenal hyperplasia – omega flap vaginoplasty – clitoroplasty – androgen insensitivity syndrome – feminizing genitoplasty

Paul N., Adhikari S. Experience with pediatric and adult cases of ambiguous genitalia reconstructed with a single stage feminizing genitoplasty procedure. Acta Chir Plast 2025; 67(1): 6–12.

Introduction

Disorders of sexual development (DSD) are congenital conditions characterized by atypical development of chromosomal, gonadal or anatomic sex resulting in ambiguous genitalia. Disorders of sexual development can arise from a varied set of causes. The term DSD encompasses any congenital condition present at birth where the genitalia is atypical in relation to the chromosomes or gonads. The karyotype serves as a prefix to classify DSDs. These conditions are intricate and rare [1,2].

During the neonatal stage, when an infant is born with ambiguous genitalia reflecting a possible diagnosis of DSD, there is a need to provide the family with not only the gender assignment but also a specific diagnosis of the condition [2,3].

The most common reasons for ambiguous genitalia in newborns, comprising more than 50% of all cases, are congenital adrenal hyperplasia (CAH) and mixed gonadal dysgenesis. The global occurrence of CAH and mixed gonadal dysgenesis is 1 : 15,000 and 1 : 10,000, respectively, although it varies significantly among different populations and countries [4]. DSD nomenclature categorizes these etiologies into groups which includes 46XX DSD, 46XY DSD, DSD related to sex chromosomes, XX or XY disorder of gonadal development, XY persistent Müllerian duct syndrome and malformation syndrome. [5] In patients presenting with DSD, there is usually a spectrum of disorders with the most common abnormality being enlargement of the clitoris and varying extent of fusion of the labio-scrotal folds, leading to a clinical presentation of ambiguous genitalia after birth. The urogenital sinus (UGS) may be elongated and is often located abnormally on the perineum or phallus. The vagina and urethra open into the urogenital sinus rather than separately on the perineum with the confluence being either low or high. Additionally, there is an absence of labia

minora. These factors result in formation of ambiguous genitalia. The point where the vagina meets the urogenital sinus is a set distance away from the bladder neck. The majority of urethral lengthening occurs mainly distal to the vaginal opening with increasing masculinization, which is known as urogenital sinus lengthening [6].

Among cases of genital ambiguity with a 46 XX karyotype, the most common diagnosis is CAH. Androgen insensitivity syndrome (AIS) is another significant condition resulting from androgen receptor (AR) dysfunction and a 46 XY karyotype, representing a paradigm of clinical disorder [7,8]. CAH or the adrenogenital syndrome is a group of autosomal recessive disorders encompassing enzyme deficiencies in the adrenal steroidogenesis pathway that lead to impaired cortisol biosynthesis. The most frequent cause is steroid 21-hydroxylase deficiency, accounting for more than 90% of cases. This interruption leads to an overproduction of byproducts of steroid metabolism by the adrenal, as a result of feedback via the pituitary. The severity and type of steroid block determines the different changes in glucocorticoid, mineralocorticoid, and sex steroid production that would need hormone replacement therapy for patients [9,10]. The range of symptoms can include neonatal salt wasting and atypical genitalia, as well as hirsutism and irregular menses in adult patients. The androgenic steroids cause varying degrees of masculinization in a female fetus [10]. The diagnosis in a female infant with the adrenogenital syndrome is confirmed by the presence of elevated ketosteroid levels in the urine or increased serum 17-hydroxyprogesterone. Phallic enlargement, fusion of the urethral folds forming a phallic urethra, and the level of entry of the vagina into the urogenital sinus are observed to varying degrees. Additionally, these children lack labia minora, as described above [8].

The complete form of AIS is a disorder of hormone resistance in which an individual with an XY karyotype and testes producing age-appropriate normal concentrations of androgens, is characterized by a normal female phenotype. Pathogenesis of this condition results from mutations in the X-linked androgen receptor gene, which encodes for the ligand-activated androgen receptor – a transcription factor and member of the nuclear receptor superfamily. Partial AIS refers to a phenotype of varying degrees of masculinization of the external genitalia due to partially responsive androgen receptors. Individuals with mild AIS may exhibit signs such as adolescent gynecomastia or fertility issues later in life, and this condition is known to occur in healthy men and boys. Thus, AIS can be defined as a disorder resulting from complete or partial resistance to the biological actions of androgens in an XY individual with normal testes development and production of age-appropriate androgen levels [11].

The objective of surgical reconstruction in patients presenting with ambiguous genitalia is the separation of the urinary and genital tracts allowing for normal voiding, creation of an adequate vaginal introitus and achievement of a near-normal appearance of the external genitalia. Keeping this in mind, surgical interventions should aim to achieve the following goals: (1) removal of the corpora while retaining the glans and its innervation to construct a clitoris with normal sensation; (2) reconstruction of the labia minora from phallic skin and foreskin to create a visually normal introitus; (3) vaginoplasty to establish an adequate vaginal opening onto the perineum [8].

This study takes into account a procedure of single stage feminizing genitoplasty that has been performed to achieve the abovementioned goals.

Materials and methods

From April 2021 to May 2023, a study was carried out in the Department of Plastic and Reconstructive Surgery of our hospital, in which 23 patients who underwent a single stage feminizing genitoplasty procedure for ambiguous genitalia were taken into account. Of these 23 patients, 17 had CAH and the rest 6 had varying degrees of AIS. In the CAH group, four patients were of the salt wasting type. All the patients were referred to the Department of Plastic Surgery following complete evaluation of gender, and chromosomal and biochemical data by endocrinologists. All the patients were raised as girls. It might be pertinent to mention at this stage that pediatric patients with 46 XY DSD and varying degrees of AIS were first referred to the Department of Pediatric Surgery where removal of abdominal and undescended testes were done and subsequently these patients were referred to the Department of Plastic Surgery. Nine patients were in the age group < 12 years, 9 patients were in the age range 12–18 years and 5 patients were aged > 18 years.

Apart from an ultrasound examination of the genital region and abdomen, no other radiographic assessment of the introitus was done. A proper clinical examination was done in all patients and the procedure planned according to the site of confluence. Of these 23 patients, 21 patients had a low confluence while the rest 2 had a high confluence. These patients underwent reduction clitoroplasty with glans preservation, omega flap vaginoplasty and labioplasty in a single stage. The patients were closely monitored during the postoperative period. Regular dressing and surgical site inspection were done while in hospital. For patients with AIS and a blind ending vagina, instructions were given for progressive vaginal dilatation with the option of vaginal lengthening deferred till adulthood.

The patients were then followed up at regular intervals after discharge and the outcome was analyzed based on general appearance, symmetry of labia, position of clitoris, and position of introitus.

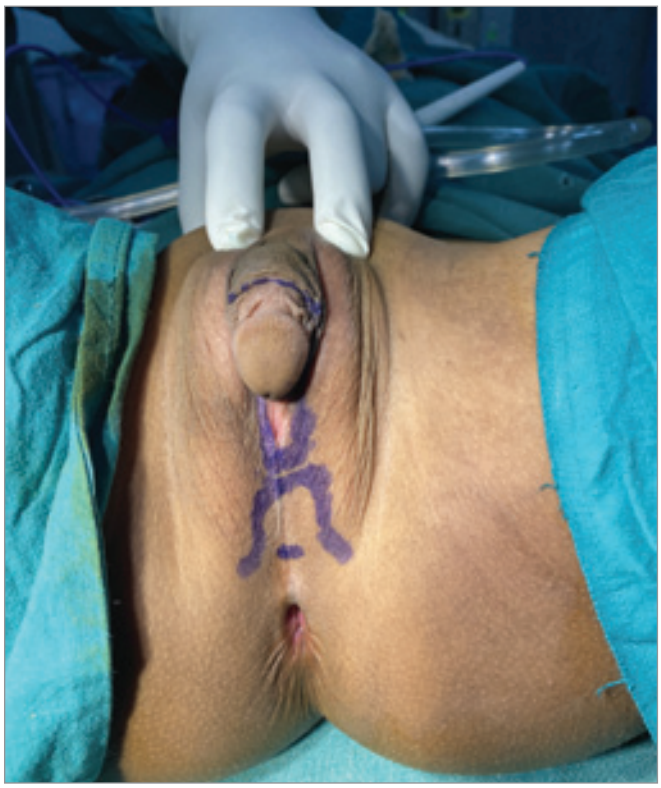

Fig. 1a. Preoperative picture with marking in a case of congenital adrenal hyperplasia with ambiguous genitalia.

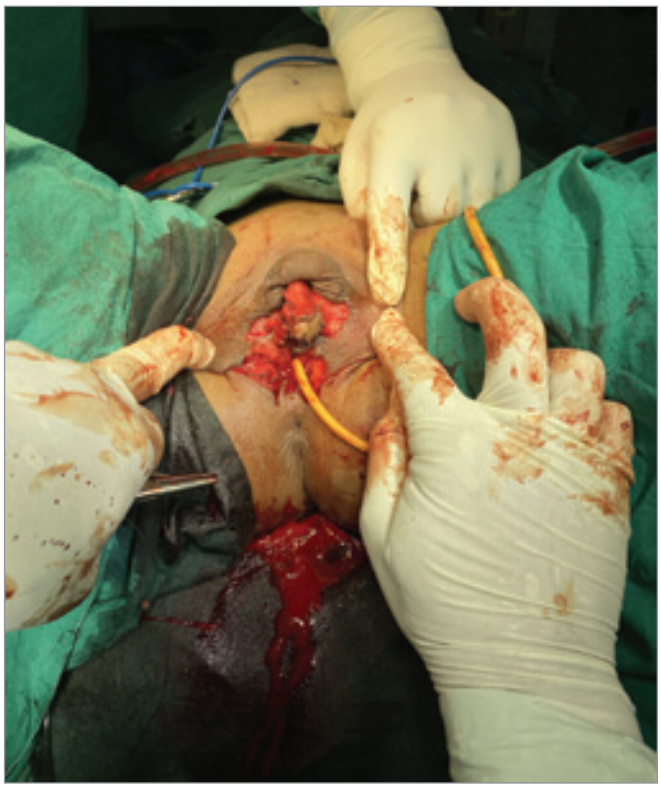

Fig. 1b. Intraoperative picture of the same case of congenital adrenal hyperplasia with ambiguous genitalia.

Senior author's technique (SA)

The patient is positioned in a lithotomy position. The procedure is begun with marking (Fig. 1a) and then a Foleys catheter is inserted into UGS. An injection of 2% lidocaine with 1 : 200,000 adrenaline is injected into the marked incision sites. The first step involves degloving of the virilized clitoris. The incision is begun at the dorsum of virilized clitoris for degloving, keeping a small part of the inner preputial skin, which would be later used in creation of the clitoral hood. The clitoral skin is then mobilized from the corporal bodies in the subdartos avascular plane dorsally up to the pubic symphysis while the dorsal neurovascular bundles are identified and carefully preserved. The degloving continues preserving UGS on the ventral aspect. Once UGS is separated from the clitoral shaft skin, attention is then directed to the raising of omega flap. The omega flap is raised in the perineal region keeping a moderate thickness of underlying fat to preserve its vascularity and this is separated from the labia majora and the posterior part of UGS (Fig. 1b). Stay sutures are then placed on either side of UGS and it is then mobilized from the corporal bodies up to the level of the bifurcation of the corporal bodies and the pubourethral ligament is identified. In cases of high confluence, the pubourethral ligament might need to be transected to achieve total urogenital mobilization, whereas in low confluence, the dissection stops at the level of the pubourethral ligament. Thereafter, two parallel incisions are marked on the ventral side of corporal bodies on each side till the level of bifurcation. The erectile tissue on both sides is mobilized and excised at the level of the bifurcation, preserving the underlying tunica and the dorsal neurovascular bundle. A rim of erectile tissue is left attached to the tunica to prevent inadvertent injury to the dorsal neurovascular bundle. The glans is preserved in continuity with neurovascular bundle. An assessment of the glans is then made. In

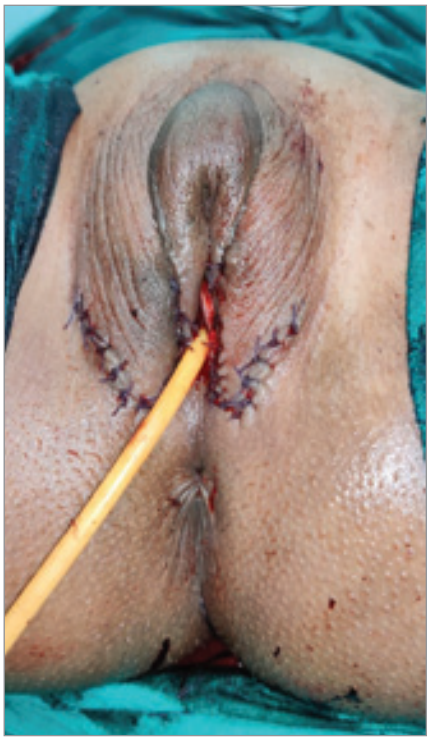

Fig. 1c. Postoperative picture of the same case of congenital adrenal hyperplasia with ambiguous genitalia.

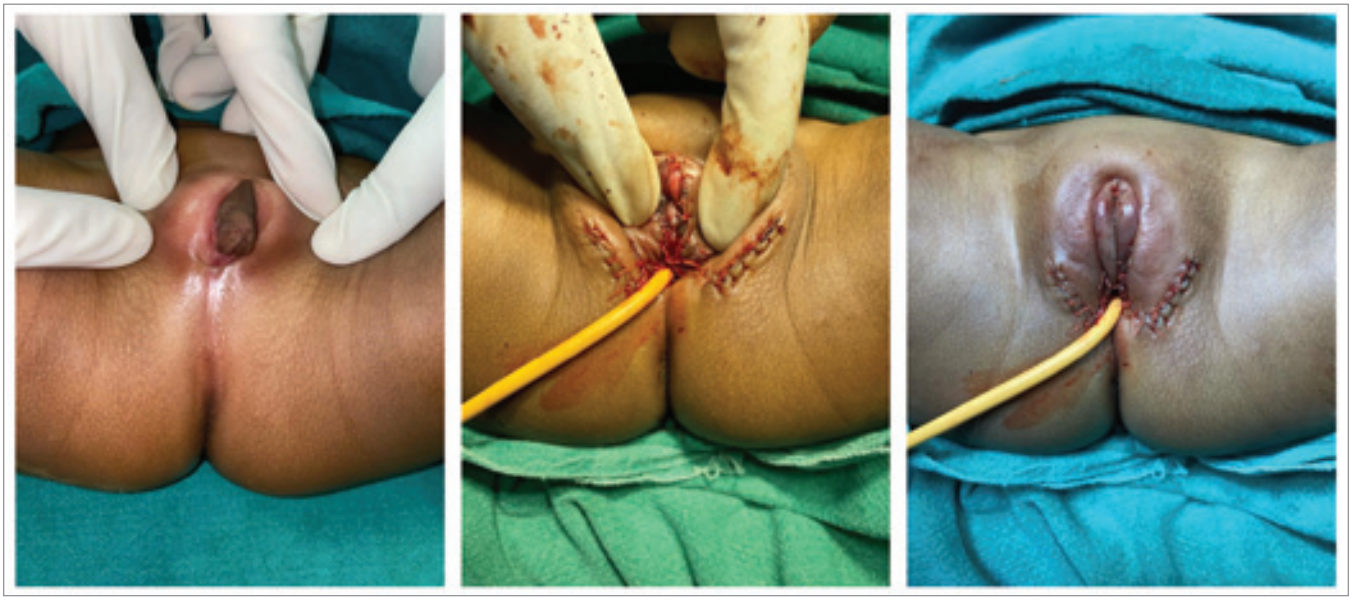

Fig. 2. Preoperative and postoperative pictures showing another case of congenital adrenal hyperplasia with ambiguous genitalia.

patients with oversized glans, an inverted V-shaped resection of the ventral part of the glans is carried out to achieve a normal clitoral shape. The glans is then fixed to the ligated corporeal stumps using plication sutures. With the glans now fixed in its position, the tunics are then next fixed to the pubic symphysis with sutures placed carefully to prevent an injury to the neurovascular bundles. The fatty tissue in the lateral part is then mobilized and used to cover the tunics to mimic a mons pubis.

The mobilized UGS is now entirely free anteriorly. The sinus is further dissected posteriorly. The sinus is now opened and the incision extended ventrally until the entire sinus is open. The vaginal opening is identified. A retractor is inserted in the vagina and further dissection continues posteriorly along the avascular layer between the two layers of Denonvilliers fascia (white plane) of vagina to mobilize the vagina. Adequate mobilization is done especially in cases of high confluence to achieve total urogenital mobilization.

The dorsal portion of the sinus is then opened in the midline to create a mucosa lined vestibule. The splitting of UGS continues till the base of the clitoris is reached, at which point UGS is secured to the clitoral base. At this point, the previously inserted Foley's catheter in UGS is inserted into the bladder. The medial edges of the sinus are sutured to the clitoral hood. The posterior omega flap is now sutured to the posterior wall of vagina, excising any redundant portion. Thereafter, the clitoral skin is used to create the labia minora. The skin is split in the midline. The previously created clitoral hood is now sewn to the clitoral skin which is additionally plicated and the medial edges of the split clitoral skin are then sewn along the lateral margins of the open urogenital sinus to create the labia minora. The distal ends of the split clitoral skin flaps are sutured to the lateral walls of the open vagina.

The V-shaped incisions are now placed at the inferior border of the labia majora and these are mobilized in a V-Y fashion to reach the inferior aspect of the vagina. The dog ears created in the medial margins are excised. The lateral aspect of the labia minora is then sutured to the medial aspect of the labia majora on each side. Mild compressive dressing is applied but we do not use any drains (Fig. 1c).

In adult patients having testes in the inguinal canal or below, a consent for orchiectomy was previously taken and these were additionally removed during the procedure.

Polyglactin 4-0 suture is used throughout the procedure (Fig. 2–5).

Results

This study included 23 consecutive cases of ambiguous genitalia who underwent this procedure of single stage feminizing genitoplasty. Of these 23 patients, 17 had been diagnosed of CAH and the remaining 6 had different degrees of AIS. Their age at surgery ranged from 4 to 23 years (mean age 14) and all of them were raised as girls. There were no significant intraoperative complications. The mean operating time was around 120 to 150 minutes and the average hospitalization period was 7–8 days. All of them received antibiotics for 7 days postoperatively. Genital edema was a common side effect observed during the postoperative period, which resolved spontaneously within 7–10 days. Minor wound disruption over the V-Y advancement flap occurred in a single case that healed adequately with secondary intention.

All the patients were followed up monthly till one year. All the cases had intact glans and normal micturition. At

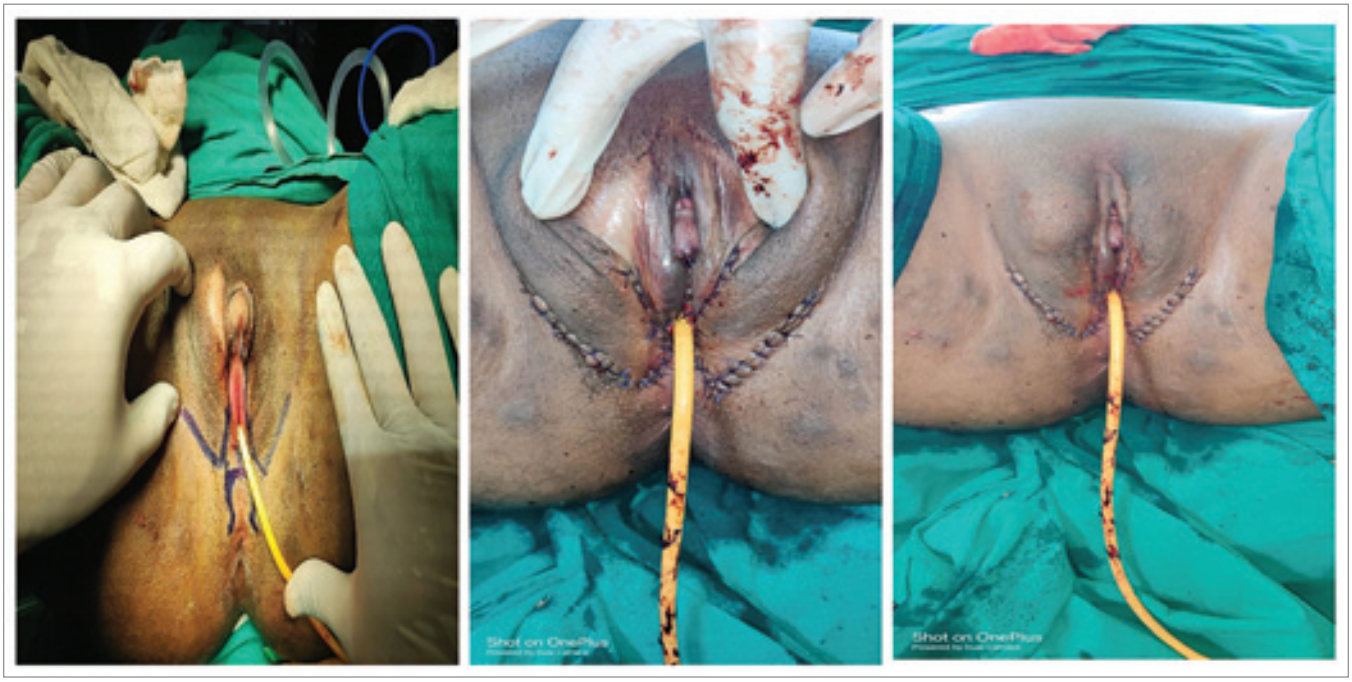

Fig. 3. Preoperative and postoperative pictures of a case of androgen insensitivity syndrome with ambiguous genitalia.

follow-up evaluation, no major complications were observed. In all the cases, the vaginal introitus was in the physiological position and was of varying size and elastic. The symmetry of bilateral labia was maintained. The parents as well as the patients were extremely satisfied with the outcomes.

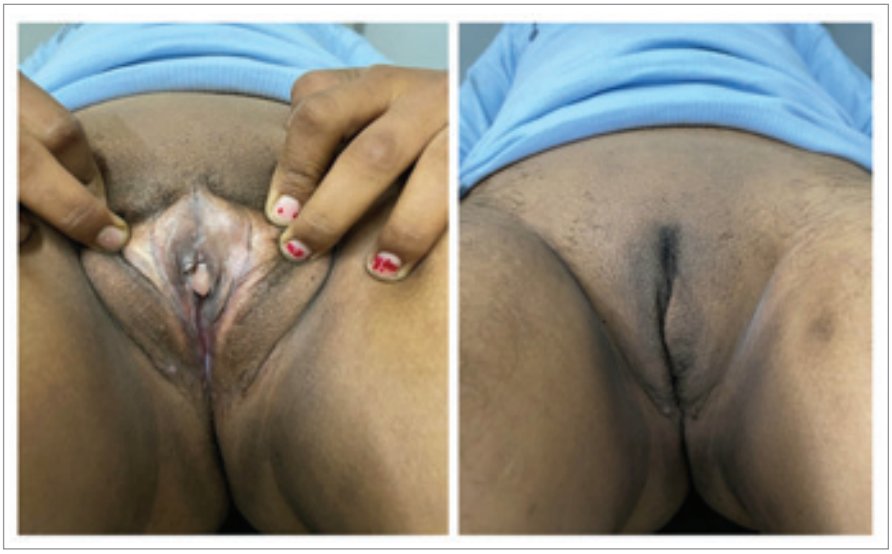

Fig. 4. The patient shown in Fig. 3 at a one-month follow-up.

Discussion

In the newborn, gender is ascribed by features of the external genitalia and whenever this is abnormal, the neonate is classified as having ambiguous genitalia. Although Anne-Fausto Sterling [12] suggested that the prevalence of intersex might be as high as 1.7%, the true prevalence might actually be around 0.018% when a more precise definition is applied [13].

The optimal gender policy is based on the fact that establishment of socially acquired gender identity generally develops by the age of 2 years and therefore early gender assignment surgery is recommended to promote optimal psychosocial and psychosexual functioning, though this has been challenged by some [14]. However, this is not always achievable in a developing nation like India because of the lack of universal newborn screening programs as well as the lack of awareness in parents [15,16].

CAH is a major cause of ambiguous genitalia. Nowadays, there are several surgical techniques available for reconstruction of ambiguous genitalia. The goal of surgical reconstruction (feminizing genitoplasty) is to separate the urinary and genital tracts to enable normal micturition, create a suitable vaginal opening, remove erectile tissue from the phallus while maintaining the neurovascular supply to the glans, prevent urinary tract complications, and most importantly, achieve a near to normal appearance of the external genitalia.

In the past, different surgical procedures have been performed to fix these anomalies and create a more typical fe-

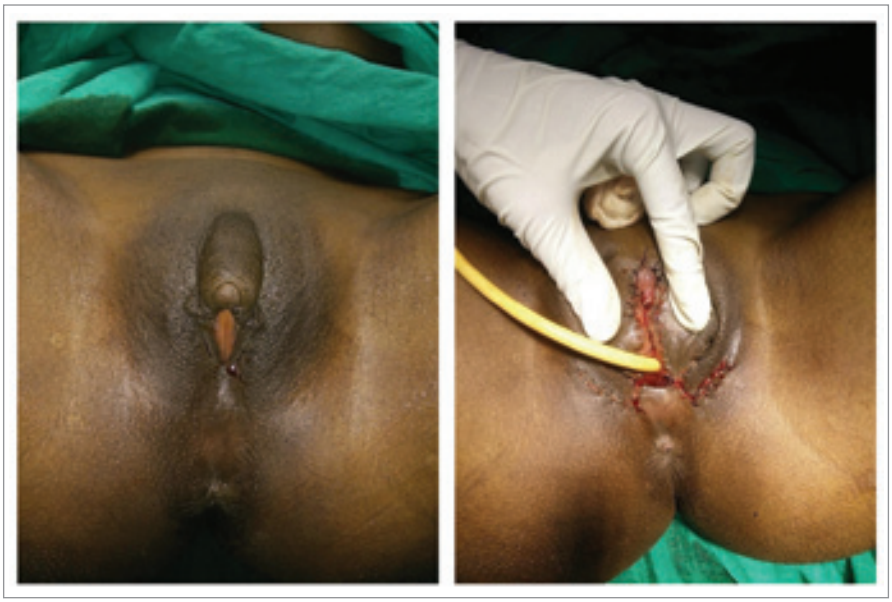

Fig. 5. Preoperative and postoperative pictures of another case of congenital adrenal hyperplasia with ambiguous genitalia.

male appearance. The initial approach to addressing phallic enlargement involved clitoral amputation [17]. This method was later reconsidered due to concerns that it was overly ablative, leading to the introduction of clitoral recession. Vaginal reconstruction has evolved toward four basic procedures: (1) cut-back vaginoplasty used in simple labial fusion; (2) flap vaginoplasty; (3) pull-through vaginoplasty, thought to be applicable to the high suprasphincteric confluence; (4) complete vaginal replacement used in those with an absent or rudimentary vagina [17,18].

Glassberg and Laugani proposed the removal of albuginea wedges to allow for folding and shortening of the corpora. The drawbacks of these methods may not be obvious until puberty, when sexual arousal can result in painful swelling of the recessed corporeal bodies. An alternative method has been created to preserve the glans, which is analogous to the clitoris, while excising the corpora. Initially, this was done by resecting the entire corpora without preserving the nerve supply to the glans [8]. In recent years, there has been more focus on preserving the nerve supply of the glans via the dorsal nerve of the phallus, a terminal branch of the pudendal nerve. Removal of the corporeal bodies with preservation of these nerves results in a cosmetic reduction in the size of the phallus while preserving the sensation of glans clitoris [8,17]. Passerini-Glazel also developed a new one-stage procedure of clitorovaginoplasty for severely masculinized female pseudohermaphrodites [19].

Our technique of genitoplasty combines modifications of previously reported feminization procedures. The primary objective of any management strategy is to establish a structure that facilitates the development of the affected patient, whether a child, adolescent or adult, into a psychologically balanced and well-adjusted individual who is content with and identifies with the gender assigned [20].

While standard protocols have emphasized the importance of early diagnosis, determining sex assignment, and performing necessary surgery during infancy, some authors have argued in recent years that such surgeries are damaging or disfiguring. They believe that since these surgeries are primarily for cosmetic purposes, they should only be carried out when the patient can provide fully informed consent [21]. Surgical reconstruction for female individuals with atypical genitalia involves decreasing the size of the clitoris and carrying out a vaginoplasty. While most experts concur that a large clitoris should be reduced in size at an early stage of life, there is a debate regarding the appropriate age for performing vaginoplasty. Simple reduction methods for vaginoplasty are sufficient for minor degrees of masculinization, but in more severe cases, these methods can result in the urethral opening being positioned in an unfavorable location within the vagina (female hypospadias). Hendren and Crawford described a technique of vaginal pull-through to address this problem of urinary incontinence, which may be the result when cutback techniques are used for severe cases of masculinization in which the vagina joins the urogenital sinus at or above the level of the urethral sphincter [18,22].

In the past, a two-stage procedure was typically used for genital reconstruction in CAH. The first stage involved the removal or reduction of the clitoris during the neonatal period, while the vaginoplasty was delayed until the child was older. Since 1989, various alterations to the traditional Hendren and Crawford technique have gained popularity and supplanted the original approach to feminizing genitoplasty. It has been observed by many that performing clitoral reduction and vaginoplasty in two stages often results in the loss of the prepuce, which is valuable tissue for reconstructing the anterior vaginal wall. To determine the appropriate type of vaginoplasty to use, it is crucial to differentiate between children with a high vaginal opening connected to the urogenital sinus and those whose vagina is low enough to be accessed by a perineal flap [8,23].

Vaginal stenosis due to fibrosis following vaginoplasty has been men-

tioned by Passerini-Glazel [23]. Such complications have not been experienced in our patients. Our patients who have reached puberty have normal appearance of external genitalia and clitoris along with adequate vaginal opening and normal micturition. In our study, all the patients underwent reduction clitoroplasty with glans preservation, omega flap vaginoplasty and labioplasty in a single stage. In all the cases, the vaginal introitus was located in the physiological position and was of varying size and elastic. The symmetry of the bilateral labia was maintained. The parents were extremely satisfied with the outcome.

Conclusion

This method of feminizing genitoplasty enables nearly all children presenting with ambiguous genitalia to undergo a one-stage reconstruction early in life, with good cosmetic results. It results in the creation of a spacious vaginal opening and a typical female vulva appearance, and the overall anatomical and functional outcomes have been outstanding. Additionally, this procedure can be carried out on individuals across all age groups, including infants.

Conflicts of interest

The authors describe no conflicts of interest.

Financial support

The authors declare that there was no financial or nonfinancial support obtained during the preparation of this article.

Declaration

All the procedures performed in this study involving human participants were in accordance with ethical standards of the institutional and/or national research committee and with the Helsinki declaration and its later amendments or comparable ethical standards.

Roles of authors

Neelanjana Paul – surgical assistant, compilation of manuscript; Souvik Adhikari – main surgeon and guide.

References

1. Kim KS., Kim J. Disorders of sex development. *Korean J Urol.* 2012, 53(1): 1–8.
2. Allen L. Disorders of sexual development. *Obstet Gynecol Clin North Am*. 2009, 36(1): 25–45.
3. Thyen U., Lanz K., Holterhus PM., et al. Epidemiology and initial management of ambiguous genitalia at birth in Germany. *Horm Res.* 2006, 66(4): 195–203.
4. Pang SY., Wallace MA., Hofman L., et al. Worldwide experience in newborn screening for classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Pediatrics.* 1988, 81(6): 866–874.
5. Witchel SF. Disorders of sex development. *Best Pract Res Clin Obstet Gynaecol.* 2018, 48: 90–102.
6. Ganesan A., Smith GH., Broome K., et al. Congenital adrenal hyperplasia: preliminary observations of the urethra in 9 cases. *J Urol.* 2002, 167(1): 275–279.
7. Allen L. Disorders of sexual development. *Obstet Gynecol Clin.* 2009, 36(1): 25–45.
8. Synder HM., Retik AB., Bauer SB., et al. Feminizing genitoplasty: a synthesis. *J Urol.* 1983, 129(5): 1024–1026.
9. Speiser PW., White PC. Congenital adrenal hyperplasia. *N Engl J Med.* 2003, 349(8): 776–788.
10. El-Maouche D., Arlt W., Merke DP. Congenital adrenal hyperplasia. *Lancet.* 2017, 390(10108): 2194–2210.
11. Hughes IA., Davies JD., Bunch TI., et al. Androgen insensitivity syndrome. *Lancet.* 2012, 380(9851): 1419–1428.
12. Fausto-Sterling A. Sexing the body: gender politics and the construction of sexuality. *New York: Basic Books* 2000.
13. Sax L. How common is intersex? A response to Anne Fausto-Sterling. *J Sex Res.* 2002, 39(3): 174–178.
14. Slijper FM., Drop SL., Molenaar JC., et al. Long-term psychological evaluation of intersex children. *Arch Sex Behav.* 1998, 27(2): 125–144.
15. Ammini AC., Gupta R., Kapoor A., et al. Etiology, clinical profile, gender identity and long term follow up of patients with ambiguous genitalia in India. J Pediatr Endocrinol Metab. 2002, 15(4): 423–430.
16. Rajendran R., Hariharan S. Profile of intersex children in South India. *Indian Pediatr.* 1995, 32(6): 666–671.
17. Rink RC., Adams MC. Feminizing genitoplasty: state of the art. *World J Urol.* 1998, 16(3): 212–218.
18. Hendren WH., Crawford JD. Adrenogenital syndrome: the anatomy of the anomaly and its repair. *Some new concepts. J Ped Surg.* 1969, 4(1): 49–58.
19. Passerini-Glazel G. A new 1-stage procedure for clitorovaginoplasty in severely masculinized female pseudohermaphrodites. *J Urol.* 1989, 142(2): 565–568.
20. Göllü G., Yıldız RV., Bingol-Kologlu M., et al. Ambiguous genitalia: an overview of 17 years' experience. *J Pediatr Surg.* 2007, 42(5): 840–844.
21. Kipnis K., Diamond M. Pediatric ethics and the surgical assignment of sex. *J Clin Ethics.* 1998, 9(4): 398–410.
22. Gonzalez R., Fernandes ET. Single-stage feminization genitoplasty. *J Urol.* 1990, 143(4): 776–778.
23. Farkas A., Chertin B., Hadas-Halpren I. 1-Stage feminizing genitoplasty: 8 years of experience with 49 cases. *J Urol.* 2001, 165(2): 2341–2346.

Neelanjana Paul, MD
Department of Plastic Surgery
Institute of Post Graduate Medical Education & Research
244, A.J.C Bose Road
Kolkata-700020
West Bengal, India
itsneelanjanahere@gmail.com

Submitted: 31. 10. 2024
Accepted: 20. 3. 2025