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Case Report

Ambiguous genitalia with positive reproductive outcome: a case report

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ABSTRACT

Ambiguous genitalia are an uncommon phenotypic manifestation of the urogenital system that may indicate the presence of a life-threatening underlying disease. The development of normal male or female external genitalia is a consequence of a series of genetic and physiological processes beginning with sex determination and proceeding through internal and external reproductive structure differentiation following the genesis of a zygote. While the failure to progress normally through the stages of sex determination and differentiation is referred to as a disorder of sex development (DSD), not everyone with DSD has ambiguous genitalia. As a result, it is critical to ascertain the etiology as soon as feasible when uncertainty is noticed. The purpose of this case study is to highlight the favorable reproductive result associated with ambiguous genitalia.

Keywords: Ambiguous, Genitalia, Physiological, DSD

INTRODUCTION

Ambiguous genitalia is an uncommon disorder in which an infant's external genitals do not seem male or female. The genitals of a child with ambiguous genitalia may be underdeveloped, or the child may exhibit traits of both sexes. External sex organs may differ from internal or genetic sex organs.[1]

When a newborn's genitalia are ambiguous, it indicates that the child's gender is unknown. Around 1% of all births result in kids with ambiguous genitalia, such as an abnormally large clitoris or an abnormally tiny penis. In more uncommon instances-between 0.1 and 0.2% of live births-genitalia is so unclear that medical professionals are consulted. A baby, for example, may have a vagina but not a uterus, cervix, or ovaries. A youngster may be deficient in male body characteristics-producing hormones such as testosterone. A newborn may have male and female organs, including testes and a rudimentary penis.[2] When uncertainty is noticed, it is critical to ascertain etiology as soon as possible. The development of normal male or female external genitalia results from a series of genetic and physiological processes beginning with sex determination and proceeding through internal and external reproductive structure differentiation following the genesis of a zygote. While failure to progress normally through the stages of sex determination and differentiation (for example, total androgen insensitivity syndrome) is referred to as a DSD, not all individuals with DSD have ambiguous genitalia. This chapter focuses on genital ambiguity in persons with a 46, XY or 46, XX chromosomal compliment; however, DSD, including genital ambiguity, can occur in individuals with other sex chromosome combinations such as 45, X/46 XY.[3]

Genetic sex is decided at conception and is responsible for the differentiation of the gonad. The differentiation of the gonad influences the development of both the internal and external genital tracts and phenotypic sex, which occurs later in development (about 5-6 weeks of gestation). Male and female genitalia are distinguished by the presence of similar features along the urogenital ridge. Around 4 weeks after conception, primordial germ cells move from the yolk sac wall to the mesonephros-derived urogenital ridge. Additionally, the urogenital ridge contains precursor

cells for follicular or Sertoli cells and steroid-producing theca and Leydig cells. On the vaginal ridges, the "indifferent" gonads develop (Figure 1).[4]

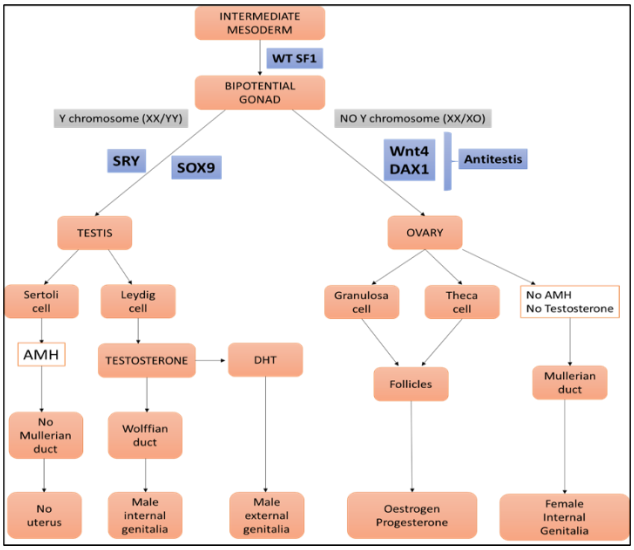

Figure 1: Sex differentiation. SRY, a sex-determining region on the Y chromosome; TDF, testis determining factor; AMH, anti-Müllerian hormone; DHT, dihydrotestosterone; WT1, Wilms' tumor suppressor gene; SF1, steroidogenic factor 1; SOX9, SRY-like HMG-box; Wnt4, Wnt=a group of secreted signaling molecules that regulate cell to cell interactions during embryogenesis; DAX1, DSS-AHC critical region on X chromosome.

CASE REPORT

A rare case of ambiguous genitalia, grown-up as female after reconstructive surgeries with the positive reproductive outcome by LSCS reported at the department of obstetrics and gynecology at Dr. M. K. Shah medical college and research centre, Chandkheda, Ahmedabad. On 25/12/21 at 2:20 pm, she gave birth to the healthy female child of birth weight 2.81 kg with no abnormality reported. The patient was under consultation of Dr. Lalit Kapadia from the period of her birth to the delivery of her baby.

Case history

She belonged to the mid-socioeconomic status and had 04 siblings. Including her, all 5 delivered by normal vaginal deliveries. No similar or any other congenital anomaly was observed in other siblings. They are normal and their offspring are also normal. Also, no accidental/ intentional drug exposure occurred during the antenatal (ANC) period of her. However, the patient's mother had a history of tuberculosis after 3rd delivery, and AKT treatment was taken for 6 months. No documented evidence is available for the same as it was almost before 30 years.

The patient was born via normal vaginal delivery (NVD) on 11th August 1989 at Gomtipur-Ahmedabad, with ambiguous genitalia, i.e.no defined sex (gender) at the time of birth and was advised to wait for further life development. The patient is born with clitoromegaly with ectopic urethral opening with a male look alike. Karyotyping on 21st June 1990 at the age of one-year favored female sex although she has grown up as a male child with all living habits of a male child including play and clothing and rest except urination.

Further, Thelarche developed was reported firstly in 2002, then after menarche was reported in 2003 and later pubarche in 2003. 1st episode of menses occurred at the age of 13 years, and it was for 3-4 days after that episode with an interval of one month patient was admitted to civil hospital, Ahmedabad, for one and a half months, and on 4th August 2003, the hormonal study was conducted, i.e., FSH:1.58, LH:6.72, testosterone: 0.52. The patient was considered as female based on the observed initial clinical symptoms. As in the local examination on April 2003, Rudimentary penis was reported along with normal labia, vagina, pubic hairs, and breast development. Then after, USG abdomen and pelvis was performed on 28th May 2003, and reports showed liver, GB, spleen, pancreas, B/L kidney as normal and uterus anteverted and 6.8×2.7×2.6 cm size with visualization of the right ovary; however, left ovary was not seen.

However, this fact was not accepted by the patient herself initially, as the patient wanted to be of the male gender. Hence, psychiatric reference and guidance were provided for gender dilemma as well as for sex identity by showing images to her at juvenile age for sexual identification and differentiation (to define male and female). Further, parental support and psychiatric counseling performed to explain future marital issues future consequences of being male gender after reconstructive surgery helped the patient to understand the situation and agreed to consider the reconstructive surgery for the female gender. Therefore, reconstructive surgery was performed for clitoromegaly with the abnormal urethral opening reconstruction. The post-operative period was uneventful, and the patient got discharged after the completion of surgery.

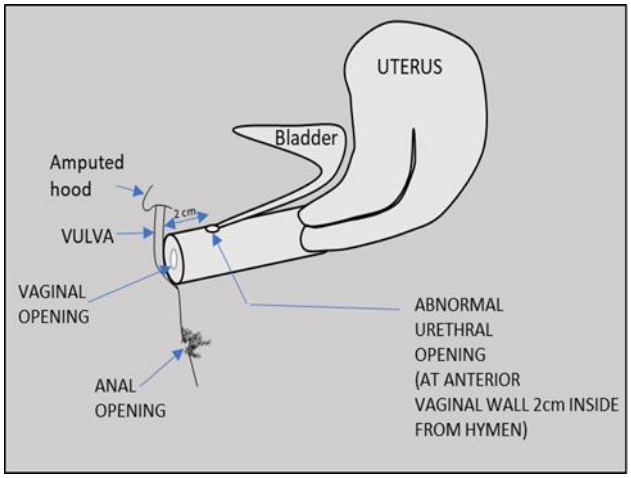

Figure 2: Anatomical diagram of uterus.

Psychological aspect during childhood

No defined problem was reported in childhood. Except she was living the life as a male child till 2002. Therefore, she had psychological trauma, fear of society, and anxiety about her actual gender revealing. Hence, she lived in fear and shame of it during childhood. Certain neighbors and a few others relatives knew about her sexual identity problem. Additionally, she did not like calling or considering herself as a girl.

Life post reconstructive surgery

The patient had complaints of urinary retention and went to the private practitioner for treatment and suprapubic catheterization done and kept for 15 days due to urethral reconstructive surgery. The patient didn't get relief and went to the civil hospital where she got operated on and routine urethral catheterization was done and SPC was removed, and the patient's condition improved and was stable. From 2003-2005 and till 2011-12, she had regular menses, and normal bowel and bladder habits with the psychological trauma of gender dilemma persisted but with the development of fully grown-up female genitalia, including adult female breast development, she accepted herself as female.

Reproductive life post reconstructive surgery

She got married to 2 years elder male on 10th Feb 2018, after a discussion of all these ambiguous genitalia-related issues. He accepted her wholeheartedly with confirmation of childbearing capacity under the consultation of Dr. Lalit Kapadia at civil hospital Ahmedabad. They faced initial difficulty in sexual intercourse due to narrow spacing. However, conservative management was suggested for vaginal spacing, i.e. By finger spacing method and vaginal mold (Acrylic mold size of 1.5-2-inch diameter with 4-inch length) on 8th May 2018.

After having 3 years of active married life, she conceived spontaneously with LMP of 9th April 2021 and passed the uneventful antenatal period. She delivered a female child of 2.81 kg by LSCS on 25/12/21 at 2:20 pm without any gross congenital anomalies. During delivery, the intra-op was uneventful, and the post-catheterization was done by smaller size 10-no. Foley's catheter. Further, the post-partum period was uneventful, and mother-baby both were stable and well. Also, she has no urinary problem and passes urine at regular intervals.

DISCUSSION

The newborn with abnormal genital development presents a difficult diagnostic and treatment challenge for the gynecologist providing care. A definitive diagnosis must be determined as quickly as possible so that the appropriate treatment plan can be established to minimize medical, psychological, and social complications.[5] The purpose of this study was to provide an extensive review of the clinical characteristics of a patient with ambiguous genitalia and later on with positive reproductive output, in the department of gynecology, Gomtipur hospital, and Ahmedabad.

The genetic sex of a baby is determined at conception based on the presence or absence of sex chromosomes. The X chromosome is found in the mother's egg, while the X or Y chromosome is found in the father's sperm, depending on the father's gender. If the father's X chromosome is present in the newborn, it is a genetic girl (two X chromosomes). A genetic man is defined as a child who receives the Y chromosome from his father (one X and one Y chromosome). The sex organs of both males and females grow from the same tissue. The presence or absence of male hormones and the number of chromosomes in this tissue determine whether or not it develops into male or female organs.[1]

In some cases, a disruption of the steps that define sex can result in a mismatch between the external genital appearance and the internal sex organs, or a discrepancy between genetic sex and the external genital appearance (XX or XY). Genetic male fetuses that are born without or with insufficient levels of male hormones have ambiguous genitalia. In contrast, genetic females who are born with inadequate or no levels of male hormones have the same condition. Gene-related changes in prenatal sex development and the formation of ambiguous genitalia have been linked to mutations in particular genes. Abnormal chromosomal anomalies, such as the absence or presence of an additional sex chromosome, can also result in the development of ambiguous genitalia. Some cases of ambiguous genitalia may be challenging to diagnose because the source is unknown.[1]

Infertility and an increased risk of cancer are two of the most serious consequences of ambiguous genitalia. The ability to bear children for persons who have ambiguous genitalia is dependent on the precise diagnosis. Those who are genetically predisposed to congenital adrenal hyperplasia, for example, may generally become pregnant if they want to do so. Specific tumors are at increased risk as a result. There is a link between particular problems of sexual development and an increased risk of certain forms of cancer.[5,6] However, our study mainly focuses on ambiguous genitalia with positive reproductive output. The scope of the ambiguous genitalia problem is not minor. An inappropriate approach to this problem poses an undue risk to the integrity of the physical and psychosexual health in the future for these children.

Highlights

Physical examination

Clitoral enlargement with B/L labial enlargement with fusion and with vaginal opening with the abnormal urethral opening.

USG

Ultrasound was performed on 28/5/2003, for abdomen and pelvis: liver, GB, spleen, pancreas, and B/L kidney were normal, and uterus anteverted, i.e., 6.8×2.7×2.6 cm size with visualization of right ovary and left ovary not seen

Hormonal examination

On 4/8/2003 hormonal study was performed, and FSH:1.58, LH:6.72, testosterone: 0.52 were recorded.

Reconstructive surgery

In Nov/Dec 2003, reconstructive surgery with vaginoplasty was performed. The clitoral hood was removed with abnormal urethral opening, i.e., Labial separation, to create vaginal space during surgery.

CONCLUSION

Having ambiguous genitalia is not a medical condition; instead, it is a developmental abnormality of sex development. Oblique genitalia are usually visible at or shortly after delivery, and it can be quite stressful for parents and children. The medical team will investigate the reason for ambiguous genitalia and give information along with counselling that can assist the family in making decisions about the baby's gender and any further required treatment. It is crucial to have a decent knowledge about DSD to analyze parents' attitudes and provide appropriate counselling. Management of DSD needs consistent classification, adequate cytogenetic facilities, and a customized approach along with integrated team management. At present, patients with ambiguous genitalia stand a far better chance of receiving a rapid diagnosis, appropriate replacement therapy, and functional surgical reconstruction than before.

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