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Ambiguous genitalia–A social dilemma in Bangladesh: A case report

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ABSTRACT

INTRODUCTION: Ambiguous Genitalia is a form of birth defect in which sex of the newborn cannot be readily distinguishable because of atypical appearance of the external genitalia.
CASE REPORT–CLINICAL FINDINGS: The patient, an 8 months old baby, was identified as a female baby since birth; but, some senior members of their neighborhood raised confusion regarding sex differentiation of the child. On examination, there was enlarged clitoris or micropenis, bifid scrotum or labioscrotal fold, and separate presence of urethral and vaginal orifices. Right testis was found in the middle of right inguinal canal and left testis was found near the deep inguinal ring.
CASE REPORT–DIAGNOSIS, THERAPEUTIC INTERVENTION AND OUTCOME: The ultrasonogram showed a rudimentary uterus measuring 26.7 × 7.27 mm with no ovary and the right testis was found in the mid inguinal canal while the left testis was in the deep inguinal ring. Diagnostic endoscopic procedure showed normal appearing testes in deep inguinal ring, about 2 cm vaginal remnant with normal urethra and bladder. Hormonal study revealed low serum testosterone with normal DHT and high oestradiol while the karyotyping revealed 46XY. After 3 months of hormonal treatment, multiple surgical interventions were taken to correctly determine the sex of the child. Later on the patient was discharged with follow up advices.
CONCLUSION: 'Ambiguous genitalia' is considered as a taboo in Bangladesh; hence, the social awareness regarding this curable disorder is a timely need in Bangladesh.

1. Introduction

Ambiguous genitalia may be a consequence of a disorder of sexual development resulting in a defect at birth where the external genitals do not have a typical resemblance to that of a boy or a girl [1]. The common causes of ambiguous genitalia in genetic females are congenital adrenal hyperplasia (CAH), maternal ingestion of substances with male hormone activity during pregnancy, male hormone producing tumour in female foetus or mother, whereas possible causes in genetic males are Leydig cell aplasia, 5-alpha-reductase deficiency, androgen insensitivity syndrome, and maternal ingestion of substances with female hormone activity during pregnancy [2,3]. Congenital Adrenal Hyperplasia seems to be more common in people of Hispanic, Slavic, Italian and European Jewish ancestry which is followed by mixed gonadal dysgenesis (MGD) [3]. The incidence of genital ambiguity is 1 per 4500, though some degree of male under-virilisation or female virilisation may be present in 2% of live births [4,5]. A study showed that the incidence is about 1 in 18,000 births in UK [3]. There have been insufficient data regarding incidences of ambiguous genitalia or other similar birth defects in the low and middle income countries (LMICs) including Bangladesh. Most cases of ambiguous genitalia can be diagnosed at birth by equivocal features of external genitalia [6]. The outcome of the treatment of ambiguous genitalia has been reported differently in different studies. A study showed that most of the participants were content with their body image. 90% of men and 83% of women had sexual experience with a partner. Both men and women were satisfied with their sexual function. [7]. For majority of cases of extreme genital ambiguity in 46,XY individuals, both male or female sex of rearing could lead to successful long-term outcome. [7]. However, another study showed that there has been unsuccessful sexual life of the treated patients [8]. A table of the literature reports on DSD with title, author and journal details is provided in Table 1 [7–18].

But, most of these reports are mainly from the western world. The literature review suggests that there is insufficient data regarding ambiguous genitalia from LMICs. This article portrays a report of a case of ambiguous genitalia which was managed at a tertiary level public health facility of Bangladesh. In this article, the case study has been reported according to the SCARE criteria [20].

2. Case report

In May 2014, an 8 months old baby hailing from a remote district of Bangladesh presented with the complaints of difficulty in identification of sex by the parents for the previous 02 months.

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Table 1
Literature reports on DSD and similar cases.

Authors	Title	Reference details
Georgette Beatriz De Paula et al.	408 Cases of Genital Ambiguity Followed by Single Multidisciplinary Team during 23 Years: Etiologic Diagnosis and Sex of Rearing	International Journal of Endocrinology [11]
V. Anantha Kumari, A. Vani	Ambiguous genitalia: a clinical and chromosomal study	International Journal of Research in Medical Sciences [14]
Carol AB Warren	Gender reassignment surgery in the 18th century: A case study	Sexualities [17]
Thyen U. et al	Epidemiology and Initial Management of Ambiguous Genitalia at Birth in Germany	Hormone Research in Paediatrics [16]
Claude J. Migeon et al.	Ambiguous Genitalia With Perineo-scrotal Hypospadias in 46,XY Individuals: Long-Term Medical, Surgical, and Psychosexual Outcome	Paediatrics [7]
Hazel Glenn Beh, Milton Diamond	An Emerging Ethical and Medical Dilemma: Should Physicians Perform Sex Assignment Surgery on Infants with Ambiguous Genitalia?	Michigan Journal of Gender & Law [19]
Milton Diamond	Paediatric management of ambiguous and traumatized genitalia	The Journal of Urology [12]
William George Reiner	Assignment of sex in neonates with ambiguous-genitalia	Current Opinion in Paediatrics [15]
Alice Domurat Dreger	"Ambiguous Sex"—or Ambivalent Medicine?: Ethical Issues in the Treatment of Intersexuality	The Hastings Center Report [18]

According to the statement of the parents, the baby was delivered at home and they had identified their child as a female baby since birth. But, within the previous two months of reporting at this hospital, some senior members of their neighborhood raised confusion regarding sex differentiation of the child which ultimately drove them to the department of Paediatric Surgery of a tertiary medical college hospital for proper advice which is about 250 kilometers away from their residence. On examination, there was enlarged clitoris or micropenis, bifid scrotum or labioscrotal fold, and separate presence of urethral and vaginal orifices. Right testis was found in the middle of right inguinal canal and left testis was found near the deep inguinal ring. The ultrasonogram showed a rudimentary uterus measuring 26.7 × 7.27 mm with no ovary and the right testis was found in the mid inguinal canal and left testis was in the deep inguinal ring. Diagnostic endoscopic procedure showed normal appearing testes in deep inguinal ring, about 2 cm vaginal remnant with normal urethra and bladder. Hormonal study revealed low serum testosterone with normal DHT and high oestradiol while the karyotyping revealed 46XY.

According to the decision of medical board comprising experts from different fields Testosterone was given in injectable form for consecutive 3 months as a part of multiphasic intervention plan and parents were counselled and convinced to convert her into male sex. After the hormonal treatment first stage of surgical intervention was performed during which vaginal remnant was found to be capacious measuring more than 5 cm. Right testis was found to be softer in consistency and smaller in size. Biopsy was taken from the same gonad and orchiopexy was performed. In the second setting, seperation and closure of vaginal orifice was performed after having the biopsy report which conferred that this testicular tissue have degenrative changes. During this phase chordee was corrected as well followed by placement of buccal mucosal strip for urethral plate. In the third and final setting of surgical intervention urethroplasty, left orchiopexy and scrotal reconstruction were performed. Later on the patient was discharged with follow up advices to contact endocrinologist.

3. Discussion

3.1. Standard treatment of DSD and its multidisciplinary treatment in the study settings

Infants born with the disease represent a neonatal medical emergency for physical, social and psychological reasons and demand a multidisciplinary team for the management [4,21,22]. The team must include aptly trained paediatric surgeons or urologist, paediatric endocrinologist or neonatologists with their support staff along with backing of geneticists, psychologists, ethicists, biochemists, counsellors and gynaecologists. [21,23]. The tertiary level hospitals (commonly known as medical college hospitals) are therefore required to manage the cases of DSD. In Bangladesh, currently, there are 31 medical colleges which have such mix of health care providers, although the genre of geneticists is usually scarce in the country. Accurate diagnosis, gender assignment, medical and surgical treatment and sharing of medical information with parents and patients are four key issues for management of ambiguous genitalia [24]. The proper diagnostic procedures should exhibit external appearance, internal anatomy, genetic make-up, and hormonal profile [6]. History has been of paramount importance to establish the diagnosis and start the management [2,6,25]. In the case presented in this article, history was taken quite carefully. Some of the key issues during the child birth could not be explored because of the delayed presentation of the baby. There might have been recall bias as well regarding extracting history from the patient's parents. Initial investigations determine whether the child is an under-virilised male or a virilised female from which differential diagnoses are established and further investigations planned [4]. As the differential diagnosis and relative investigations will depend on the genetic sex, an urgent karyotype has to be carried out within earliest possible time [4]. A more conservative approach has been followed considering the economic condition of the patient although extensive investigations are advised to reach the definitive diagnosis by different scientific papers [2,22,24–26]. Minimum diagnostic tests with high sensitivity and specificity were performed in this case to attain the correct diagnosis. It is noteworthy to mention that the sample for karyotyping was sent to a laboratory situated in New Delhi, India since in Bangladesh, during that period, the service for karyotyping was not available. However, currently the investigation is performed in the only medical university of Bangladesh. The treatment constituted both the available ones i.e. hormonal therapy followed by surgical measures although the timing of the following steps such as gender assignment and surgery is still a topic of debate [21]. Gender assignment should be aimed at offering the best prospect for healthy puberty and sex life [1,6,22,24] and should be attempted after comprehensive clinical, genetic and biochemical investigation actively involving the parents with detailed discussion and explanation [6,16,22,25]. The consensus for the management of ambiguous genitalia suggests that if it is decided to assign male gender to the infants, 10–25 mg of testosterone enanthate or cypionate is given intramuscularly for once a month for consecutive 3 months to ensure that the penis responses to androgens or to improve the size of penis prior to surgery [27]. However, patients with 5-alpha reductase deficiency should receive dihydrotestosterone replacement. In case of unavailability, higer dose of testosterence can be given as a replacement to overcome the enzyme block [27].