VOLUME - 9, ISSUE - 7, JULY - 2020 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Pathology

MIXED GONADAL DYSGENESIS PRESENTING AS PENILE HYPOSPADIAS- A CASE REPORT

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ABSTRACT The Mixed Gonadal Dysgenesis includes congenital anomalies including cytogenetic, gonadal and genital alterations.1 Hypospadias is recognized as a pediatric urological disorder, the incidence of which is on the rise over the last two decades. This is the second most common congenital anomaly seen. The incidence was 1 in 250 or 300 cases two decades back. However recent data suggest an increase of 1 in 50 cases in the recent years.2 This anomaly has been associated with undescended testis in about 5% of the population.3 Does it all seem so simple or is there anything lurking behind these features. Disorders of sexual differentiation a rare scenario however can present a hypospadias.

KEYWORDS :

INTRODUCTION

Disorders of sexual differentiation are congenital condition within which the development of chromosomal, gonadal and phenotypic sex is atypical.4 Disorders of sexual differentiation are a rare problem. It is a medical emergency as often the parents reaction to birth of child with atypical genitalia is one of shock and concern. The worry often being to which pole the child should be reared to. One of the less worrying presentations is penile hypospadias.5

Case history

4 year old male presented to pediatric surgery with penoscrotal hypospadiasis and perineal fistula. He is a full term normal delivery baby with a birth weight of 3.5kgs and milestones achieved. On ultrasonography atrophic uterus with fallopian tube and atrophic gonad was observed. Karyotyping of the patient was done and analysis revealed 46XY chromosome. We received specimen in two pieces, uterus measuring 2x1x0.5cm, cut section shoed endometrial cavity and other tissue is an irregular pale brown soft tissue measuring 2x1x1cm. Histology showed rudimentary uterus, streak ovary, and fallopian tubes and epididydimal tissue. On correlating with clinical, radiological and histology a diagnosis of mixed gonadal dysgenesis was made.

DISCUSSION

DSD are group of conditions which were earlier referred to as intersex. Ambiguous genitalia is the most common presentation of DSD, which often put the parents in a dire state regarding sexual development of the child. According Chicago DSD6 were broadly classified into four categories such as

1. Sex chromosome DSD
2. 46XX DSD
3. 46XY DSD
4. Others.

We are presenting a case from sex chromosome DSD. Mixed gonadal dysgenesis now classified under sex chromosome DSD, is a condition of unusual and asymmetrical gonadal development leading to an unassigned sex differentiation. MGD represents an intermediate between pure gonadal dysgenesis and OT-DSD. 7 A number of differences have been reported in the karyotype, most commonly seen are a mosaicism 45, X/ 46, XY, similar to ours. The external genitalia, internal genitalia, and gonadal phenotype are highly variable. 46XY genotype can be difficult to distinguish from 46XY OT-DSD. This mosaicism may cause formation of a dysgenetic or malformed gonad. This is referred to as a streak gonad that does not produce enough testosterone.8 Ambiguous genitalia is the most common presentation for individuals with a 45X/46XY karyotype accounting for about 60% of involved individuals. In clinical examination, undescended testis (cryptorchidism) and/or hypospadias may be found. Any patient with penoscrotal hypospadias and undescended testis, karyotyping is necessary.9 Structural rearrangement of the Y chromosome were seen in 63% of MGD patients.10 In our case, mosaic chromosomal pattern with a chromosome marker showed SRY mutation upon proposal of MGD as the primary diagnosis. Formation of the testis from the undifferentiated embryonic gonad depends on the presence of the short arm of the Y chromosome, containing SRY-sequences. Testosterone production stimulates development of the Wolffian system and induces male development of the external genitalia, failing which, differentiation proceeds along female lines and Müllerian structures are formed.11 Undifferentiated gonad to become a testis, a minimal amount of SRY is required. These cases require complete clinical examination, cytogenetic and histopathological findings, and the performance of new diagnostic methods such as screening of micro deletions in sexual chromosomes.

CONCLUSION

Often insufficient care of physical and psychology of children can be avoided with good, prompt medical evaluation and genetic counseling commencing from neonatal period.Every case of penile hypospadias must be evaluated to rule out any underlying DSD. Early diagnosis and evaluation is very important for preventing psychological sequels as well as ruling out other abnormalities.

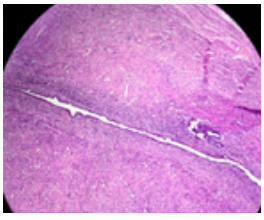

Figure 1- low power view of endometrium

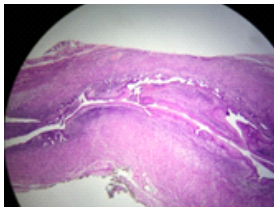

Figure 2- low power view of endometrium and cervix

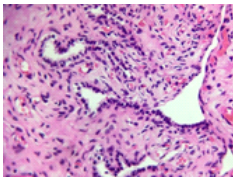

Figure 3- epididymal tissue

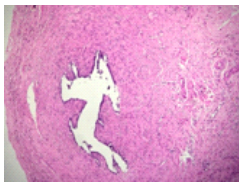

Figure 4- tubal epithelium

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