Case Report

Disordered sexual development: a case report

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Abstract

We are reporting a case of disorder sexual development where on the basis of external genitalia it was difficult to decide the gender of such person. After local and general examination, biochemical tests, ultrasound examination, and karyotyping the chances of male gender were more. Such cases are difficult to deal looking at the psychological aspect of diagnosis for subject and parents. Such cases need to be managed by a multispecialty team including psychiatrists or psychologists.

KEY WORDS: Disordered sexual development, external genitalia, karyotyping, hypogonadotrophic hypogonadism

Introduction

Disorders of sexual development (DSDs) are important clinical situations associated with mental trauma for the parents. DSDs terms are applicable for the condition in which sex is congenitally abnormal because of chromosomal, gonadal, or anatomical reasons.[1] In one out of 4500 births, the external genitalia are found to be abnormal and cause difficulty in deciding the gender of the child.[1] For such children it is important to diagnose the condition thoroughly before coming to the conclusion about the management. Management of such cases should be done by multidisciplinary team of pediatric endocrinologist, psychiatrist/psychologist, pediatric urologists, geneticists, and may also include social worker and nurse.[2] Here we report a case of DSD, which highlights the importance of multidisciplinary approach for the management of such cases.

Case Report

This child was brought to our outpatient department (OPD) by the parents with the following complaints. The child had not attained menarche so far. The mother noted swellings on either side of the urethral orifice, which were gradually increasing in size. The child had already been taken to a nearby hospital soon after the birth, because the external genitalia appeared ambiguous. But the local doctor had reassured them and advised no specific treatment. At the age of 7 years, the child was again taken to a medical college hospital as the genital ambiguity was more evident then. This time also they were reassured and no definite treatment or surgical intervention was advised and hence the patient was reared as a female child. There was no history of long-term drug therapy and the patient had no visual problems or any discharge from nipple. The other sibling is male of 12 years with normal development and sexuality. In addition, there is no positive history of sexuality problems in any of the family members. The patient is moderately built and nourished. No pallor, icterus, cyanosis, or clubbing. Height of patient was 146 cm, weight was 41.1 kg, BMI was 20.45 kg/m^2, and arm span was 163 cm. Upper and lower limbs were proportionate to the body. Thyroid appeared to be normal. There was normal interspace between two nipples. Vitals were normal. In the case of secondary sexual characteristics breast and nipples were male pattern; there was no axillary, chest, or facial hair. Pubic hair was male pattern. On examination of external genitalia micropenis was noted with penoscrotal hypospadias. There was a bifid scrotal swelling with palpable tissue suggestive of testis. There was no vaginal orifice. Prostate was not palpable on per rectal examination. Systematic examination was normal. There was no abnormality in routine blood examination. On ultrasound examination there was no evidence of uterus and ovaries and normal testis noted on both sides with normal size and echo texture. There were no renal or suprarenal anomalies. There was no

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barr body found on buccal smear examination. On hormonal evaluation serum prolactin was found to be 4.27/mL (within normal limits), follicle stimulating hormone 12.05 mIU/mL, luteinizing hormone 8.07 mIU/mL, growth hormone 11.521 ng/mL (normal limit), and serum testosterone 5.35 ng/mL (male range 1.8–9 ng/mL). Thyroid profile was normal. Karyotyping was done and 46 XY pattern was observed. On X-ray, normal ossification centers were observed.

Discussion

In the case of primary amenorrhea the most important causes are Mullerian agenesis, gonadal dysgenesis, and hypogonadotropic hypogonadism.[3] Karyotyping should done to evaluate the presence of Y chromosome as it may indicate the chance of malignant transformations. In the case of 46, XY gonadal agenesis chances of malignant transformation are 20%–30%. In the case of hypogonadotrophic hypogonadism the streak of tissue should be removed if Y chromosome is found in karyotyping as it may be transformed to malignancy.[4]

Management of such conditions needs multifaceted approach including etiological diagnosis, assignment of gender, genital surgery, and disclosure of information about the condition to person and parents. This requires efforts from the multiple clinicians including psychiatrists. There may be immense psychological trauma because of the condition and that may affect the quality of life of the patient.[5] As management of such conditions require multispecialty team, so patient should be taken/referred to the tertiary-level health center without wasting much time in lower centers.

The case reported here is consistent with the 46, XY undervirilized subject. There may be various causes including somerare causes may be considered for the etiology of such situation, such as 17-hydroxysteroid dehydrogenase mutations, androgen insensitivity syndrome, 3β-hydroxysteroid dehydrogenase mutations, 5α-reductase deficiency, ovotesticular DSD, gonadal dysgenesis, and single gene mutations related to testicular development. It is observed in that in such cases exact etiology could not be found in around half of the cases.[2] Even in the case of a definite diagnosis genetical and biochemical pictures are many a times found to be very complex creating problem in exact diagnosis. In this case karyotyping shows 46, XY and external examination revealed micropenis and ultrasound examination revealed both testis. There was no uterus and ovary on ultrasound examination. These features indicate the male sex of this subject. But previous studies reported that such subjects are usually not satisfied with the gender assigned to them after examinations. There are cases where 46, XY people established with female gender identity.[6] Contrary is also true where 46, XY people who were initially assigned with female gender later changed it to male gender.[7,8]

Conclusion

This was a rare case of DSD where subject was found to be 46, XY on karyotyping. Local and general examination was consistent with male gender. This case highlights the complex etiology, diagnosis, and management of such cases. These cases should be managed as early as possible preferably at tertiary care centers.

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