

A case of ambiguous genitalia diagnosed in a young male presenting clinically with left side obstructed inguinal hernia

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Abstract

Ambiguous genitalia is a disorder of sex development (DSD). It occurs when something goes wrong during pregnancy to interrupt or disturb the fetus developing sex organs. Presence or absence of male hormones control the development of sex organs. A disruption of steps that determine sex can result in a mismatch between external genitals and internal sex organs or the chromosomal sex (xx or xy). Other causes can be mutations in certain genes or chromosomal abnormalities like missing sex chromosome or an extra one. Ambiguous genitalia is usually not life threatening, but can create social problem for the child and family.

Keywords: ambiguous genitalia, inguinal hernia.

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INTRODUCTION

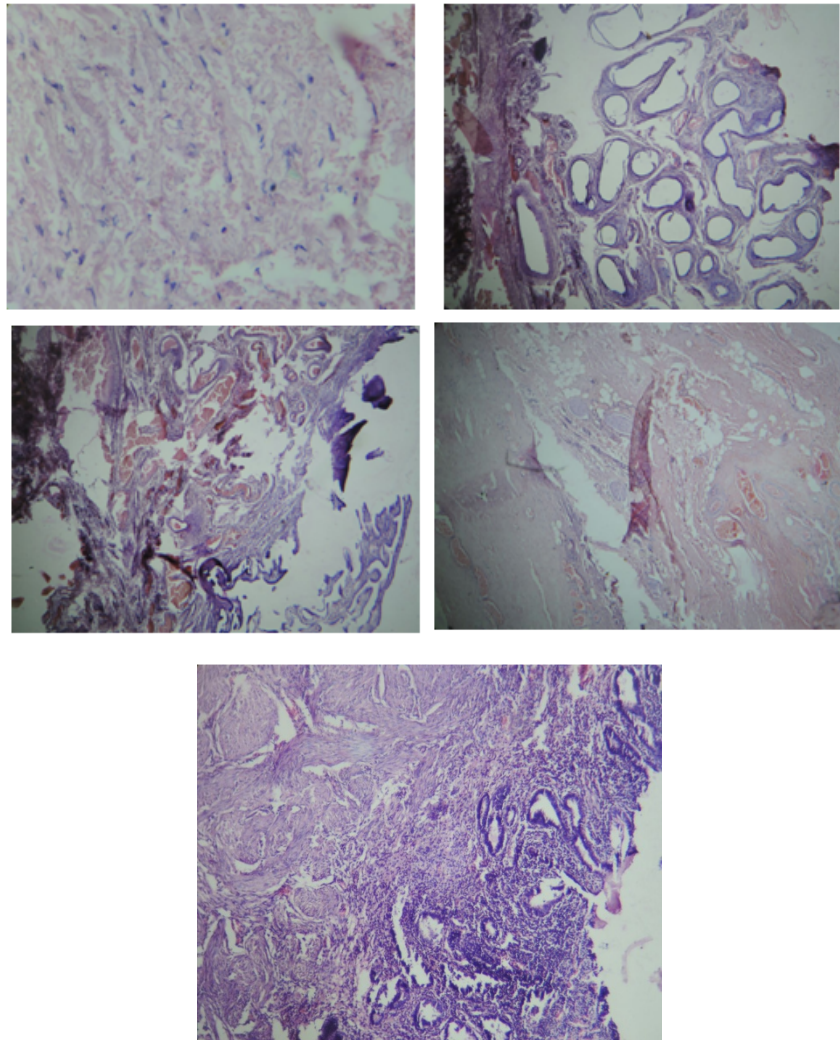
Disorder of sex development (DSD) include a heterogeneous group of heritable disorder of sex determination and differentiation. This may include chromosomal or monogenic disorder which inhibit or change primary genetic or endocrine pathways of normal sex development¹. However in many cases no definitive cause could be found. Such cases represent an enormous challenge for the clinician. Male and female sex organs develop from same tissue whether this tissue becomes male organ or female organs depends on the chromosomes. In males, a region of Y chromosome triggers development of testicles which produce male hormones controls the development of sex organs.

Mismatch between the external genital and internal sex organs or the chromosomal sex (xx or xy).

CASE REPORT

A 24 year old male presented with left sided inguinal swelling along with left sided obstructed inguinal hernia. He had history of inguinal swelling since 10 days and undescended testes since birth. On physical examination we came to know a left sided obstructed inguinal hernia with an absence of left testes. Further exploration of the inguinal canal shows an indirect inguinal hernia containing a left omental tissue along with atrophic testes, omentum and? vas deferens or fallopian tube, along with a solid mass to which a tube is attached? vas deferens or fallopian tube which in turn is attached to prostate by a thick band of fibrous tissue. Thus complete surgery was performed by left herniorrhaphy and also total excision of the solid mass with attached structures. A biopsy was also taken from the left testes. The histopathological examination revealed normal structure of epididymis, vasa deferentia and uterus and fallopian tubes without ovaries. Post operative karyotype analysis indicated a normal male genotype 46XY and further semen analysis shows asthenoterato spermia.

Histopathology of female reproductive organs in the same patient



Ambiguous genitalia is a birth defect where the genitals do not have the typical appearance of either a boy or girl¹. It is not a life threatening problem but can cause social problems for child and family. It is usually obvious at or shortly after birth. Ambiguous genitalia is a disorder of sex development.⁵

Possible cause in genetic females;

- CAH(3,4)
- Prenatal exposure to male hormones

Possible cause in genetic males;

- Impaired testicle development
- CAH
- Androgen insensitivity syndrome
- Abnormalities with testes or testosterone
- 5 α – reductase deficiency.

Others can be pseudohermaphroditism, true hermaphroditism, mixed gonadal dysgenesis (MGD).

Risk factors

Family history of infertility, genital abnormalities, CAH, abnormal physical development during puberty, unexplained deaths in early infancy.

Test and dx

Usually diagnosed at birth or shortly after DX tests include hormonal levels, chromosomal analysis, USG pelvis and abdomen, x-ray or minimally invasive surgery to collect sample of reproductive organs.

Treatment

The goal of treatment is to ensure sexual function and fertility as well as long term psychological and social well being.

Medications: Hormones, Surgery: To preserve normal sexual function, create more natural looking genitals.

CONCLUSION

Ambiguous genitalia can cause social problem for the child and family. A team of experienced specialists, including neonatologist, geneticists, endocrinologists, psychiatrists or social workers should be involved in child's care. Appropriate diagnosis and approach is most important for the future of child.

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