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Ambigious Genitalia

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Abstract

Disorders of sex development (DSD) include a hetero-geneous group of heritable disorders of sex determination and differentiation. This includes chromosomal aswell as monogenic disorders, which inhibit or changeprimarily genetic or endocrine pathways of normal sex development. However, in many patients affected, nodefinitive cause for the disorder can be found. Therefore,the birth of a child with ambiguous genitalia stillrepresents an enormous challenge. For the structuringof diagnostic procedures, decision making and alsotherapeutic interventions, a highly specialised team of physicians of different subspecialties and experts for psychosocial care is needed to counsel parents andpatients accordingly. This article focuses on the genetic and molecular origins of DSD, the new DSD nomenclature, the consecutive classification, and steps fordiagnosis. Furthermore, we discuss the approach to the family for comprehensive counselling in decisionmaking for their child

Keywords: Ambiguous genitalia; Chromosome; Congenital heart disease; Pseudohermaphroditism.

Introduction

Ambiguous genitalia appear as a large clitoris or small penis. Because there is variation in all of the processes of the development of the sex organs, a child can be born with a sexual anatomy that is typically female, or feminine in appearance with a larger-than-average clitoris (clitoral hypertrophy), or typically male, masculine in appearance with a smaller-than-average penis that is open along the underside. The appearance may be quite ambiguous, describable as female genitals with a very large clitoris and partially fused labia, or as male genitals with a very small penis, completely open along the midline ("hypospadic"), and empty scrotum.

Fertility is variable. According to some,[1,2] the distinctions "male pseudohermaphrodite", "female pseudohermaphrodite" and especially "true hermaphrodite"[3] are vestiges of outdated 19th century thinking. According to others, the terms "male pseudohermaphrodite", and "female pseudohermaphrodite" are used to define the gender in terms of the histology (microscopic appearance) of the gonads.[4]

Case Summary

5 days male baby was admitted in our NICU with low birth weight (birth weight -2.06 kgs) with ambigious genitalia with undescended testis with ectopic anal opening at penoscrotal junction. He is 38 weeks 1day by gestational age extracted by emergency LSCS due to h/o of oligohydroamnios to mother with IUGR baby on antenatal USG with meconium stained liquor in "SHOW". Baby cried immediately after birth ,cry was good and birth weight is 2060 gms .Our case is 2th issue of non consanguinous marriage his parents.

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First conceptus was spontaneous abortion at 4month of gestational age.

On examination, her pulse was 136 beats/ minute & respiratory rate of 34 cycles / min Neonatal reflexes are normal. On examination baby has ambigious genitalia with undescended testis with ectopic anal opening. Baby passes meconium from ectopic anal opening located at penoscrotal junction.

Her investigations were as follows:

Hb: 14.3 gm%

TLC: 8000/cmm

P – 55, L – 39, E – 02, M – 03, B – 01.

Platelet count: 1.49 lacs/cmm.

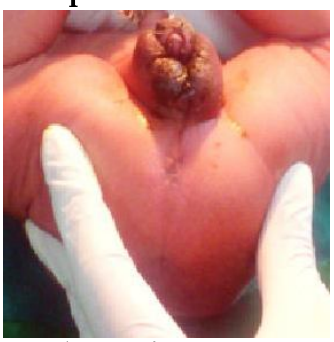
Chest x-ray is normal

USG cranium is within normal limit.

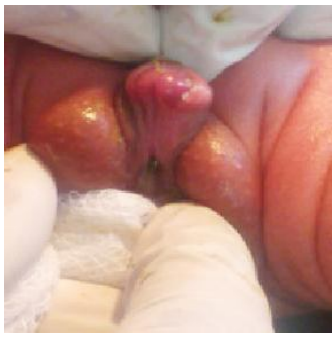
USG abdo pelvis shows low placed right kidney with non-visualization of left kidney. No visualization of testis bilaterally.

2D ECHO & Color Doppler of heart states presence of patent ductus arteriosus of 1.5 mm with left to right flow & patent foramen ovale 4 mm with left to right shunt. No coarctation of aorta no pulmonary hypertension S/O acyanotic heart disease.

Imperforate anus



Ectopic anal opening at penoscrotal junction



Discussion

Ambiguous genitalia (also known as a typical genitalia) is a birth defect (or birth variation) of the sex organs that makes it unclear whether an affectednewborn is a girl or boy. This condition occurs approximately once in every 4500 births. The baby seems to have a mixture of both female and male parts -for example, they may have both a vulva and testicles. Associated intersex conditions form alebabies include hypospadias, where the urethral opening is located in an unusual position such as the underside of the penis.[5]

The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals. Tests (including ultrasound, x-rays and blood tests) are needed before the baby's sex can be identified. Mild forms of ambiguous genitalia may be characterised by a large (penis-like)clitoris in baby girls or undescended testicles in boys.[6]

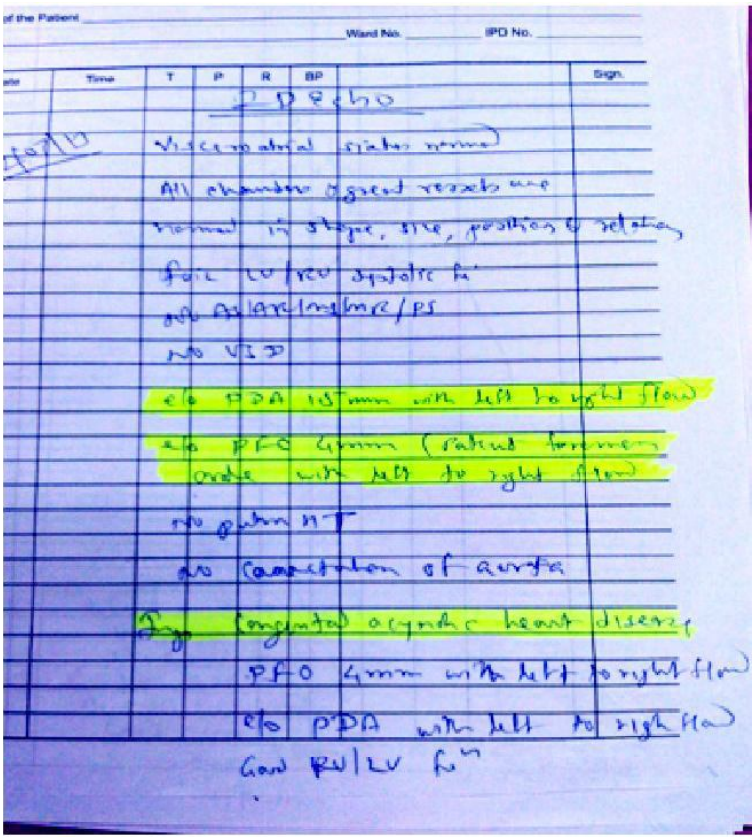
Sexual Determination During Embryo Development

A baby's sex is decided at conception. The mother's egg provides an X chromosome and father's sperm determines the baby's sex by contributing either an X or Y sex chromosome. An XX embryo is female while an XY embryo is male. Both female and male embryos develop in exactly the same way and have identical gonads and genital parts until around the eighth week of gestation. The sexual determination process includes:

Girls

The internal genital parts transform into the uterus, fallopian tubes and vagina. The gonads turn into ovaries which start producing female sex hormones. The lack of male hormones is fundamental in allowing the development of female genitalia.

2D-ECHO report



Boys

The internal genital parts transform into the prostate gland and vas deferens. The gonads turn into testes which start producing male sex hormones. The presence of male hormones allows the penis and scrotum to develop. [7]

Different Types of Ambiguous Genitalia

The different types of ambiguous genitalia include:

- The baby has ovaries and testicles, and the external genitals are neither clearly male nor female.
- The baby has ovaries and a penis-like structure or phallus.
- The baby has undescended testes and external female genitals including a vulva.

A Range of Causes

For typical genital development, the gender ' message' must be communicated from the sex chromosomes to the gonads. The gonads must then manufacture appropriate hormones and the genital tissues and structures have to respond to these hormones. Any deviations along the way can cause ambiguous genitalia. Some specific causes include:

- Androgen insensitivity syndrome (AIS) - a genetic condition characterised by the fetal tissue's in sensitivity to male hormones. This affects genital development. For example, a newborn may have some of the female reproductive organs but also have testicles.
- Congenital adrenal hyperplasia (CAH) - an inherited condition that affects hormone production. A child with CAH lacks particular enzymes, and this deficiency triggers the excessive manufacture of male hormones. For example, female genitals are masculinised.

Birth Defects: Ambiguous Genitalia

- Sex chromosome disorders - instead of having either XX or XY sex chromosomes, a baby may have a mixture of both ('mosaic' chromosomes); or specific genes on the Y chromosome may be inactive; or one of the X chromosomes may have a tiny Y segment attached to it. Research at the University of California at Los Angeles (UCLA) indicates that ambiguous genitalia can be caused by the doubling up of a particular gene (named WNT-4) on the sex chromosome. This variation will interfere with male sexual development so that a genetically male baby will appear female.
- Maternal factors - the pregnant mother may have had an androgen - secreting tumour while pregnant, and the excess of this male hormone affected her baby's genital development. In other cases, the placenta may have lacked a particular enzyme which failed to deactivate male hormones from the baby as a result, both the mother and the female baby were masculinised by the excess of these hormones.[8]

Diagnosis Methods

There are currently no prenatal tests that can detect ambiguous genitalia. American research into the WNT-4 gene suggests that a prenatal test could one day be developed. Tests performed at birth to determine the baby's gender can take about one week and may include:

- Physical examination
- Hormone tests using blood, urine or both
- Genetic tests using blood, urine or both
- Ultrasound scan
- X-rays.

Treatment

Treatment options to help assign the baby a definite gender may include:

Parental Counselling: Successful sex assignment and identity for the child depends largely on the attitude of the parents. It is important that both the mother and father are fully informed about their child's condition. Support groups may provide help in this area.

Surgery: For example, an overly large clitoris may be trimmed, or a fused vulva separated, or undescended testicles relocated into the scrotum. However, surgical gender assignment depends heavily on what genital structures the surgeons have to work with.

The majority of babies with ambiguous genitalia have been brought up as girls. A few operations may be needed, usually begun in the child's first year. Further surgery might be required during adolescence. Some intersex support groups feel that surgery is not always the answer, particularly when the gender of the child is not clear. Others suggest that surgery should wait until the child is old enough to decide for themselves. However, most medical professionals advocate early surgical and hormonal intervention for the sake of clearly establishing the child's gender and sense of belonging in society.

Counselling for the Child: The child needs to be informed and talked to about their diagnosis in a very careful way.

Hormone Therapy: During their teenage years, the child may need hormone supplementation therapy to help bring on puberty. A child with CAH will need to have daily hormone therapy.

Possible Long-Term Problems

Some of the possible problems faced by a person born with ambiguous genitalia may include:

- Infertility
- Problems with sexual functioning.[9]

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