Birth defects - ambiguous genitalia

Ambiguous genitalia (also known as atypical genitalia) is a birth defect (or birth variation) of the sex organs that makes it unclear whether an affected newborn is a girl or boy. This condition occurs approximately once in every 4,500 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 for male babies include hypospadias, where the urethral opening is located in an unusual position such as the underside of the penis.

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.

Sexual determination during embryo development

A baby's sex is decided at conception. The mother's egg provides an X chromosome and the 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.
- **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.

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 insensitivity 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.

- **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.

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 options

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
- Feelings of insecurity and uncertainty about their gender identity, such as feeling like the opposite gender to the sex that was determined earlier in life.

Where to get help

- Your doctor
- Endocrinologist
- Genetic counsellor
- Endocrinology clinic counsellor

- Congenital Adrenal Hyperplasia Support Group Tel. (03) 5227 8405
- AIS Support Group Australia Tel. (03) 9315 8809

Things to remember

- 'Ambiguous genitalia' is a birth defect of the sex organs that makes it unclear whether an affected newborn is a girl or boy.
- Causes include genetic variations, hormonal imbalances and malformations of the fetal tissues that would have otherwise evolved into genitals.
- Treatment aims at assigning the baby a specific gender.
- Treatment options include corrective surgery, hormone therapy, peer support and counselling.

This page has been produced in consultation with, and approved by:

Murdoch Children's Research Institute

Content on this website is provided for education and information purposes only. Information about a therapy, service, product or treatment does not imply endorsement and is not intended to replace advice from your doctor or other registered health professional. Content has been prepared for Victorian residents and wider Australian audiences, and was accurate at the time of publication. Readers should note that, over time, currency and completeness of the information may change. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions.

For the latest updates and more information, visit www.betterhealth.vic.gov.au

Copyright © 1999/2012 State of Victoria. Reproduced from the Better Health Channel (www.betterhealth.vic.gov.au) at no cost with permission of the Victorian Minister for Health. Unauthorised reproduction and other uses comprised in the copyright are prohibited without permission.