



Ambiguous Genitalia

What does “ambiguous genitalia” mean?

The term “ambiguous genitalia” means that a newborn baby’s genitals look different than the genitals of most boys or girls. Parents and doctors cannot tell right away what the newborn’s sex is. For example, a baby can have an enlarged clitoris that looks more like a small penis, and fusion of the labia so that they look more like a scrotum. In some babies, the penis does not form or is very small, and the opening where urine comes out can be at the base of the penis, not at the tip. Conditions that have these features are called disorders of sex development, or DSD.

Definitions

- **Sex chromosomes:** the X and Y chromosomes. Each person usually has 46 chromosomes, including two sex chromosomes. Most females have two X chromosomes (XX). Most males have one X and one Y chromosome (XY).
- **Sex hormones:** chemicals made by the body that influence sex development and sexual function, including estrogen (a female hormone) and testosterone (a male hormone).
- **Gonads:** reproductive organs (testicles and ovaries). At puberty, the testicle makes sperm and male hormones. The ovary releases eggs and female hormones.
- **Genitals:** external sex organs, such as the penis and scrotum in males, and the clitoris and labia in females.
- **Sex assignment:** the sex in which a child is raised.
- **Gender identity:** how people think of themselves—as male or female.

How do the sex organs develop in the fetus?

Shortly after conception, the gonads typically develop into either testicles or

ovaries. In males, hormones made by the testicles then cause the penis and scrotum to form. In the absence of male hormones, the female fetus develops a clitoris, vagina, and labia.

What causes ambiguous genitalia?

There are many different causes of ambiguous genitalia, which are sometimes grouped according to the sex chromosomes present.

46, XX DSD occurs when the female fetus is exposed to excess male hormones at the time that the genitals are forming. The most common cause is congenital adrenal hyperplasia, a condition in which the adrenal glands (small glands in the abdomen) overproduce male hormones.

46, XY DSD occurs in the male fetus when:

- The testicles don’t develop properly
- The testicles can’t make enough testosterone
- The body can’t use testosterone properly.

Sex chromosome DSD occurs when there is an atypical number or combination of sex chromosomes. Causes vary according to the type of variation.

What tests will your child need?

Your child’s doctor will examine your child and ask about your family’s medical history. The doctor will check your child’s chromosomes and measure hormone levels in the blood and possibly urine. Ultrasound tests and scans can sometimes show the internal sex organs. Occasionally, doctors might examine tissue samples from the gonads or look at them with a special telescope (laparoscopy). Sometimes a diagnosis can be made in a few days; other times it takes longer.

What factors will be considered in making a sex assignment for your child?

You may be faced with a choice of whether to raise your child as a boy or a

girl. You and your doctor will consider:

- The cause of your child’s condition (if the cause can be found) and likely gender identity
- The appearance of your child’s genitals
- Options for surgery
- How well your child’s gonads are likely to function in the future
- Whether your child will be fertile
- Your family’s preferences and cultural beliefs.

What is the treatment for ambiguous genitalia?

The goals of treatment are to ensure your child’s long-term emotional well-being, sexual function, potential for fertility, and a stable gender identity. Treatment can include hormone therapy (usually at puberty) and surgery to improve sexual function and for cosmetic reasons. Doctors may recommend surgery for some patients when they are infants. In other cases, parents may choose not to pursue surgery or to delay it until the child is old enough to contribute to the decision.

What resources can help with ambiguous genitalia?

You and your child should see a team of health care providers, with specialists in newborn health, genetics, endocrinology (hormones), pediatric surgery or urology, and psychology. Many parents and children have found that support groups are helpful.

Resources

Find-an-Endocrinologist:
www.hormone.org or call
1-800-HORMONE (1-800-467-6663)

Mayo Clinic information about ambiguous genitalia: www.mayoclinic.com/health/ambiguous-genitalia/DS00668

Hormone Foundation information about CAH (see this link and search for CAH):
www.hormone.org/Resources/factsheets.cfm

Consortium on Disorders of Sex Development. Handbook for Parents:
<http://dsgguidelines.org>

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For more information on how to find an endocrinologist, download free publications, translate this fact sheet into other languages, or make a contribution to The Hormone Foundation, visit www.hormone.org or call 1-800-HORMONE (1-800-467-6663). The Hormone Foundation, the public education affiliate of The Endocrine Society (www.endo-society.org), serves as a resource for the public by promoting the prevention, treatment, and cure of hormone-related conditions. This page may be reproduced non-commercially by health care professionals and health educators to share with patients and students.

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