



Ambiguous Genitalia

WHAT DOES 'AMBIGUOUS GENITALIA' MEAN?

The term ambiguous genitalia means that a newborn baby's genitals look different from the typical genitals of either a boy or a girl. Parents and doctors cannot tell right away what the newborn's sex is.

For instance, a baby can have an enlarged clitoris (part of the female genitals) that looks more like a small penis, and fusion of the labia (the "lips" of the vagina) so that they look more like a scrotum (the pouch that contains the testicles). 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.

HOW DO THE SEX ORGANS DEVELOP IN THE FETUS?

Shortly after conception, the gonads typically develop into either testicles or ovaries. In males, the testicles make hormones that cause the penis and scrotum to form. In the absence of male hormones, the female fetus develops a clitoris, vagina, and labia.

DID YOU KNOW?

Management of ambiguous genitalia aims to ensure the child's long-term emotional well-being, future sexual function and fertility, and stable gender identity.

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.

WHAT CAUSES AMBIGUOUS GENITALIA?

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

46, XX DSD occurs when the fetus is exposed to excess male hormones at the time that the genitals form. The most common cause is congenital adrenal hyperplasia. In this condition, the adrenal glands (small glands above the kidneys) overproduce male hormones.

46, XY DSD occurs in the fetus when

- The testicles do not 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 with a blood test or by scraping the inside of your child's cheek. Also, the doctor will measure hormone levels in the blood and possibly urine. Ultrasound tests and scans can sometimes show the internal sex organs. Sometimes, doctors might examine tissue samples from the gonads or look at them with a special telescope (laparoscope).

Sometimes a diagnosis can be made in a few days; other times it takes longer.

WHAT FACTORS SHOULD YOU CONSIDER 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

HOW IS AMBIGUOUS GENITALIA MANAGED?

The goals of management are to ensure your child's long-term emotional well-being, future sexual function and potential for fertility, and stable gender identity. Treatment can include hormone therapy (most often at puberty).

Another treatment option is surgery to improve sexual function or for cosmetic reasons. Doctors may suggest surgery for some patients when they are infants. Parents also may choose not to pursue surgery or to delay it until the child is old enough to help make the decision. The choice to forego surgery is more commonly accepted today.

All surgery carries risks, so talk with your child's doctor if you are considering this option.

To help you make decisions about managing your child's condition, you and your child should see a team of health care providers. This health care team may include specialists in newborn health, genetics, endocrinology (hormones), pediatric surgery or urology, and psychology.

Questions to ask your doctor

- What is my baby's sex?
- Does my child need treatment now or later?
- Are there support groups for people dealing with this condition?
- Should my child see a pediatric endocrinologist?

RESOURCES

- Find-an-Endocrinologist: www.hormone.org or call 1-800-HORMONE (1-800-467-6663)
- Hormone Health Network information about congenital adrenal hyperplasia: www.hormone.org (search for CAH)
- Mayo Clinic information about ambiguous genitalia: www.mayoclinic.com/health/ambiguous-genitalia/DS00668
- Consortium on Disorders of Sex Development. Handbook for Parents: <http://dsdguidelines.org>
- dsd Families: www.dsdfamilies.org

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The Hormone Health Network offers free, online resources based on the most advanced clinical and scientific knowledge from The Endocrine Society (www.endo-society.org). The Network's goal is to move patients from educated to engaged, from informed to active partners in their health care. This fact sheet is also available in Spanish at www.hormone.org/Spanish.

