

Ambiguous Genitalia and Intersex Conditions

Some infants are born with “ambiguous genitalia.” This means that the visible genitals—penis and testicles or vagina and clitoris—aren’t clearly either male or female. There are many possible causes of these conditions. Expert testing is needed to determine the diagnosis, sexual identity, and best treatment for your baby.

What are ambiguous genitalia?

Ambiguous genitalia are one of many different conditions in which the outer genitals don’t clearly appear male or female. They are sometimes called “intersex” conditions.

Most babies with ambiguous genitalia have the genes of either a male or female, but with some additional characteristics of the opposite sex. Special tests, including genetic testing, are needed to determine your infant’s exact diagnosis and to decide on the best management. Infants with some intersex conditions may also have other medical problems that need treatment.

For parents, it’s upsetting to be told their newborn has ambiguous genitalia. Expert medical diagnosis and treatment and sensitive follow-up care can help to provide your baby with the best chance of a happy, healthy life.

What do they look like?

There are many causes and possible appearances of ambiguous genitalia. For many children with intersex conditions, the external genitals are not clearly male or clearly female. Infants with complete “androgen insensitivity”—in which male hormones don’t have the normal effect—look like females but are actually males.

- The genitals may look either like a small penis or an enlarged clitoris. The vagina may appear closed, resembling a scrotum (the sac containing the male testicles), or the scrotum may show some separation, resembling a vagina. Some infants have elements of both male and female genitals. Many variations are possible.
- The internal sex organs may not match the appearance of the external genitalia. For example, a baby who seems to have a penis may have ovaries, while a baby with undescended testes (male testicles that have not dropped to their normal position in the scrotum) may seem to have a vagina.
- Many infants with ambiguous genitalia are otherwise healthy, with no other medical problems. Babies with

some genetic conditions have other associated abnormalities.

What causes ambiguous genitalia?

Most cases are related to genetic (inherited) defects affecting the hormones involved in determining the appearance of the genitals (sexual characteristics). Initial examination and testing focus on finding the underlying cause of your infant’s condition.

It’s important to understand that, in every infant, the male and female genitals start out the same during early development of the baby in the womb. The development of the genitals is affected by genes and by many substances the body produces (such as hormones). That’s why so many different conditions can end up causing ambiguous genitalia: they interfere with the normal processes that determine whether the genitals will turn out to be male or female.

Some of the main causes of ambiguous genitalia are:

- *Congenital adrenal hyperplasia.* This is related to several different genetic defects that affect enzymes involved in making hormones in the adrenal gland. (The adrenal gland is a small organ near the kidney that produces a number of different hormones, including those affecting development of the genitals.) Congenital adrenal hyperplasia may cause girls to develop male characteristics or boys to develop female sexual characteristics in childhood. Some forms can cause severe illness.
- *Androgen insensitivity syndromes.* Babies with this condition are genetically male (have male sex chromosomes). However, they have a defect that doesn’t allow male hormones to properly affect the development of the genitals. Some male babies with androgen insensitivity look female, but others have more features that make them look male.
- *Sex chromosome abnormalities.* These are various genetic abnormalities in which the baby doesn’t have the normal sex chromosomes of either a male (XY) or a female (XX). These include the rare condition in which the infant is a “true hermaphrodite,” with both ovarian (female) and testicular (male) tissue.
- *Problems during pregnancy.* Some conditions in the mother during pregnancy can lead to problems in sexual development of the baby. These may include hormone-producing tumors or certain medications.

Many other causes are possible. Babies with ambiguous genitalia don’t always fall into clear categories. Expert testing and diagnosis/identification will seek to provide the best possible information on the cause of your baby’s condition.

What are some possible complications of ambiguous genitalia?

Many infants with ambiguous genitalia or intersex conditions are otherwise healthy. Others have additional medical problems that need treatment. For example, some forms of congenital adrenal hyperplasia can lead to problems with electrolyte balance and blood pressure.

- Some causes are associated with other birth defects.
- Many children with conditions causing ambiguous genitalia are unable to have children as adults. However, this is not always the case.

What puts your child at risk of ambiguous genitalia?

- Many of the genetic diseases that cause ambiguous genitalia run in families. You may carry the genes for these abnormalities without being aware of it. In many cases, however, there is no previous family history of genetic disease.
- There are some rare conditions arising during pregnancy—such as hormone-producing tumors or certain medications—that can cause ambiguous genitalia.

Can ambiguous genitalia be prevented?

Genetic counseling can help you to understand the risk of passing one of these intersex conditions on to future children. Otherwise, there is no practical way to prevent these conditions or to diagnose them before birth.

How are ambiguous genitalia diagnosed?

Examination, testing, and diagnosis are the essential first steps in determining the cause of your child's condition, finding out whether there are any accompanying medical problems, and deciding on the most appropriate treatment.

These evaluations are performed by experts in diagnosing these complex conditions, for example, a geneticist (a specialist in genetic diseases) or an endocrinologist (a specialist in gland and hormone diseases).

The results of some key tests will have a major impact on your child's diagnosis and treatment, including:

- Genetic tests. These will determine which, if any, genetic abnormality your child has. Parents and other family members may be tested as well.

- Hormone levels and other blood tests.
- Ultrasound or other tests to assess your child's genitals and internal organs of reproduction. It is especially important to determine whether ovaries or testicles are present.

How are ambiguous genitalia treated?

Your child's treatment will depend on the results of a full evaluation:

- *Hormone treatments* may be needed immediately. This is especially important for some infants with congenital adrenal hyperplasia.
- *Counseling* is an important part of treatment. The families of children with ambiguous genitalia need detailed information about their child's condition and emotional support to deal with the stress of this difficult time.
- *Sex assignment* is a difficult and sensitive issue. Although not always, sometimes a decision needs to be made as to whether the child is to be raised as a boy or a girl. Most experts feel it is important to make this decision as soon as possible after birth. Sex assignment can affect surgical issues.
- *Surgery* on the ambiguous genitalia is often required. In many situations, it is easier for the surgeon to create a functioning vagina than a functioning penis. As a result, many children with intersex conditions are raised as girls. This is obviously a difficult and emotional decision to make. Input and advice from various medical and mental health professionals can provide parents with information to help sort through all these issues.
- *Follow-up* is needed to monitor your child's growth and development. Further medical treatment may be needed as your child grows, especially around the time of puberty. Emotional and social support are important as well. With expert medical care and support, many children with intersex conditions grow up to lead normal lives.

When should I call your office?

Going through the diagnosis of an intersex condition is a very stressful time for your family. Call our office if you need help finding support or expert information regarding your child's condition.

Our office will continue to oversee your child's general health care. Call our office if you have any questions or concerns about your child's general health.