CONDITION

Ambiguous genitalia

Learn about atypical genital development in newborns and children, including possible causes, diagnostic testing, treatment options, and when urgent care is needed.

WHAT IT IS
External genitals that do not appear typically male or female

BODY SYSTEM
Reproductive and hormone systems

MAIN SYMPTOMS
Enlarged clitoris, small penis, atypical urethral opening, undescended testes

WHAT CAUSES IT
Genetic or hormonal, sometimes unknown

HOW IT IS DIAGNOSED
Physical examination, blood tests, chromosome and genetic testing

MAIN TREATMENT
Individualized hormone treatment, supportive care, sometimes surgery

CAN IT BE CURED?
Sometimes - depends on the underlying cause

IS IT AN EMERGENCY?
Sometimes - repeated vomiting or unusual sleepiness

LONG-TERM OUTLOOK
Varies by cause with long-term follow-up often needed

Overview

Ambiguous genitalia is an older medical term for external genitals that do not appear typically male or female. Health professionals may instead use atypical genitalia or describe the specific findings. These may be part of a difference of sex development (DSD), meaning that chromosomes, reproductive organs, hormones, or genital anatomy developed in an unexpected way.

Atypical genitalia is not a diagnosis. It has many possible causes, and testing does not always identify one explanation. Differences may be noticed during prenatal imaging, at birth, later in childhood, or around puberty.

Prompt assessment is important because a few causes affect salt balance, blood pressure, or hormone production. One example is congenital adrenal hyperplasia (CAH). Certain forms can

cause a newborn to lose dangerous amounts of salt and fluid.

Most evaluations should be coordinated by clinicians experienced in DSD care. Unless urgent treatment is needed, families should have time to understand the findings before making lasting decisions. Chromosomes, anatomy, fertility potential, health needs, sex assignment, and future gender identity are related but are not the same.

Symptoms

Atypical genitalia usually does not cause pain. Findings vary and may include:

Some differences appear later. A child may have puberty changes that do not match expectations, such as absent periods, limited breast development, unexpected facial hair, or a deepening voice. A groin hernia may sometimes contain a testis in a child thought to be female.

A newborn with atypical genitalia needs prompt medical evaluation even if otherwise well. Seek urgent medical care for poor feeding, repeated vomiting, unusual sleepiness, weakness, dehydration, or difficulty gaining weight. These can indicate a serious hormone or salt-balance problem.

Call emergency services if a baby is difficult to wake, collapses, has a seizure, has severe breathing difficulty, or appears seriously ill.

- A clitoris that is larger than expected
- A penis that is smaller than expected
- A urethral opening on the underside of the penis instead of at the tip
- Folds of tissue that appear partly or fully fused
- A divided or differently shaped scrotum
- One or both testes that cannot be felt in the scrotum
- Features associated with both male and female genital development

Causes and risk factors

Differences in sex development usually involve genes, hormone production, or hormone response. They are generally not caused by something a parent did during pregnancy.

Possible causes include:

Some conditions are inherited. Risk may be higher when relatives have a known DSD, CAH, unexplained infertility, unusual puberty development, or unexplained deaths in early infancy.

However, many affected babies have no family history.

- Congenital adrenal hyperplasia: CAH affects the adrenal glands. The most common form reduces cortisol production and may increase androgen hormones. Some forms also reduce aldosterone, which controls salt and fluid balance.
- Differences in androgen response: A person with XY chromosomes may produce androgens but have cells that respond partly or not at all, as in androgen insensitivity syndrome.
- Differences in hormone production: The testes or adrenal glands may not produce expected amounts of testosterone or related hormones. Examples include 5-alpha-reductase deficiency and certain enzyme deficiencies.
- Differences in gonadal development: Ovaries or testes may develop differently. Some people have both ovarian and testicular tissue, called ovotesticular DSD.
- Chromosome differences: Sex-chromosome variations, including mosaic patterns in which cells have different chromosome sets, can affect development.
- Hormone exposure during pregnancy: Rarely, an androgen-producing condition in the pregnant parent or exposure to certain hormone medicines affects fetal development.
- No identified cause: Testing may not provide a specific explanation.

Diagnosis

The first goals are to protect the child's health, understand the anatomy, and identify conditions requiring prompt treatment. The team should use respectful language and avoid repeated examinations unless medically necessary.

Evaluation often involves a pediatric endocrinologist and may include neonatologists, urologists, gynecologists, genetic specialists, psychologists, nurses, and reproductive specialists.

Clinicians ask about pregnancy, medicines or hormone exposure, family history, and previous infant illnesses or deaths. Examination assesses the genital anatomy, urinary opening, and whether gonads can be felt.

Blood tests may measure:

Newborn screening often includes CAH, but results may not be immediately available, and screening does not detect every form. Necessary testing or treatment should not be delayed while waiting for screening results.

Chromosome testing may identify XX, XY, or sex-chromosome variations. Genetic testing may

look for specific gene changes, although results can take time and may remain inconclusive.

Ultrasound may look for a uterus, gonads, or urinary tract differences. Imaging has limitations: failure to see an organ does not always mean it is absent. Imaging also cannot predict future gender identity or organ function. Selected cases may require additional imaging, hormone stimulation tests, or minimally invasive procedures.

Sex assignment discussions may consider anatomy, hormone function, likely sexual function and fertility, evidence about gender outcomes, family values, and remaining uncertainty. Families should receive balanced information and psychosocial support without pressure to decide before urgent health issues are addressed.

- Sodium, potassium, glucose, and other indicators of metabolic health
- Cortisol, aldosterone, and adrenal hormones
- Testosterone and related reproductive hormones
- 17-hydroxyprogesterone, which helps identify the most common form of CAH

Treatment and management

Treatment depends on the diagnosis and health needs. Atypical genital appearance alone does not necessarily require medical or surgical treatment.

A newborn with salt-wasting CAH may need immediate hospital care, including fluids, correction of low blood sugar or abnormal salt levels, and replacement adrenal hormones. Long-term treatment may include daily hormone medicine and a written plan for illness, injury, or surgery, when extra medication may be required.

Other hormone treatments may replace hormones the body does not make or support puberty. Choice and timing depend on the diagnosis, age, goals, and the young person's participation in decisions.

Surgery is not automatically required. It may be recommended for a blocked urinary opening, repeated infections, pain, or another clear health problem. Procedures intended mainly to change appearance or predict future sexual function require careful consideration. Risks include scarring, reduced sensation, urinary problems, repeat procedures, and results that may not match the person's later preferences.

When surgery is not medically urgent, families should ask whether it can be delayed until the child can participate in the decision. The team should explain the option of no surgery, limits of the evidence, possible effects on sensation and fertility, and the likelihood of future procedures.

Some gonads have an increased tumor risk, but the level varies by diagnosis, location, age, and gene changes. Monitoring or removal requires individualized specialist advice. Removing hormone-producing gonads may make lifelong hormone replacement necessary.

Psychological support can help families use age-appropriate explanations, protect privacy, respond to stigma, and support healthy body image. Peer-support organizations may also help.

Complications

Complications depend on the underlying condition and may include adrenal crisis, abnormal salt levels, low blood sugar, blood pressure problems, or reduced sex-hormone production.

Fertility may be reduced, although some people can have biological children, sometimes with reproductive assistance. Fertility potential cannot always be predicted in infancy.

Certain diagnoses increase the risk of urinary problems, groin hernias, or gonadal tumors. Treatment also carries risks: hormone medicines require monitoring, and surgery can affect sensation, scarring, urinary function, and future sexual health.

Secrecy, stigma, repeated examinations, unwanted disclosure, or exclusion from decisions can affect emotional health. Gender identity develops over time and cannot be determined through laboratory tests or examination. A child expressing distress about their assigned sex or body deserves supportive, nonjudgmental care.

Follow up

Follow-up is often long term. Visits may monitor growth, blood pressure, hormone levels, medication effects, puberty, urinary and bone health, and emotional well-being.

Children should gradually receive accurate, age-appropriate information about their body and diagnosis. As they mature, they should participate more fully in decisions and learn which medical details matter in an emergency.

Seek earlier medical review for new urinary problems, groin swelling, unexpected puberty changes, missed periods, medication difficulties, or concerns about growth and development.

People with adrenal insufficiency need a clinician-provided emergency plan. Vomiting may prevent oral medicine from being absorbed. Seek urgent care during significant illness, especially with repeated vomiting, severe weakness, unusual sleepiness, or dehydration.

Transition from pediatric to adult care should be planned. Adult follow-up may address hormones, sexual health, fertility, tumor-risk monitoring, mental health, and questions about

previous treatment.

Sources

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