CONDITION

Anorchia

Learn how anorchia, the congenital absence of both testicles, affects puberty, hormones, fertility, diagnosis, and long-term treatment.

WHAT IT IS
Congenital absence of both testicles

BODY SYSTEM
Reproductive system

MAIN SYMPTOMS
Neither testis felt in scrotum, small scrotum, absent puberty, infertility

WHAT CAUSES IT
Often unknown

HOW IT IS DIAGNOSED
Hormone blood tests, sometimes laparoscopy

MAIN TREATMENT
Testosterone replacement therapy

CAN IT BE CURED?
No, treatment cannot restore sperm production

IS IT AN EMERGENCY?
Sometimes, sudden abdominal, groin or scrotal pain

LONG-TERM OUTLOOK
Lifelong testosterone supports health but cannot restore fertility

Overview

Anorchia is a rare condition in which both testicles are absent at birth. The testicles, also called testes, normally make testosterone and sperm. Testosterone supports development of the male reproductive system before birth, drives many changes of puberty, and helps maintain bone, muscle, sexual, and metabolic health throughout life.

In many cases, the testes began developing during pregnancy but were lost before birth. This may happen because their blood supply was interrupted or the tissue twisted and could not survive. If the testes made hormones early enough in pregnancy, a baby may have typical male external genitalia even though no testes can be found after birth.

The timing of testicular loss affects development. Loss later in pregnancy may leave typical male genitalia and an empty or underdeveloped scrotum. Loss earlier may lead to a small penis

or genital development that does not look typically male or female.

Anorchia is different from undescended testes. With undescended testes, one or both testes are present but remain in the abdomen or groin instead of moving into the scrotum. Because an undescended testis can be difficult to locate, doctors must carefully confirm that testicular tissue is truly absent.

Complete anorchia causes testosterone deficiency and infertility unless hormone treatment is provided. Testosterone replacement can support puberty, sexual function, bone health, and general well-being, but it cannot restore sperm production when both testes are absent.

Symptoms

The first sign is usually that neither testis can be felt in the scrotum. This may be noticed during a newborn examination or later during a routine physical examination. The scrotum may appear small or less developed.

Other findings depend on when testicular function stopped before birth. They may include:

Pubic and underarm hair may still develop because the adrenal glands also make weak sex hormones. However, other testosterone-dependent changes, such as growth of the penis, deepening of the voice, and increased muscle mass, may not progress normally without treatment.

Anorchia itself does not usually cause pain. Sudden abdominal, groin, or scrotal pain requires prompt medical assessment if there is any possibility that an undescended testis is present and has twisted. Testicular torsion, or twisting that cuts off blood flow, is an emergency.

- A small penis
- An opening of the urethra that is not at the tip of the penis, known as hypospadias
- External genitalia that are not typically male or female
- Lack of expected puberty without treatment
- Low muscle mass or limited facial and body hair
- Reduced sexual desire or difficulty with erections in adults
- Infertility

Causes and risk factors

The exact cause is often unknown. One explanation is that the testes formed normally but

disappeared during fetal development after an interruption in blood flow. Possible events include twisting of the testicular blood vessels, a blood clot, or another vascular problem. These events are generally unpredictable.

A person with anorchia commonly has a 46,XY chromosome pattern. The Y chromosome usually starts testicular development. If the testes function for part of pregnancy, their hormones may guide development toward typical male internal and external anatomy before the tissue is lost.

In some people, early problems with genes involved in sex development may affect whether testicular tissue forms or survives. Genetic testing may be recommended when genital development is atypical, hormone results are unclear, or there are other health or developmental findings.

There are no well-established actions that a pregnant person could have taken to prevent most cases. Anorchia is not generally attributed to routine exercise, diet, or ordinary activities during pregnancy. A family history is uncommon, although a genetics professional may help assess recurrence concerns when a genetic cause is suspected.

Diagnosis

The main diagnostic challenge is distinguishing anorchia from bilateral undescended testes. A clinician will examine the scrotum, groin, penis, and urethral opening. The examination may also look for features that could suggest a broader difference in sex development.

Blood tests can help determine whether functioning testicular tissue is present. These may include:

The meaning of these results depends on age. Infants have a temporary period of active reproductive hormones during the first months of life. Hormone levels are naturally low during much of childhood and rise again at puberty. A pediatric endocrinologist can interpret results in the correct developmental context.

An human chorionic gonadotropin, or hCG, stimulation test may sometimes be used. This test checks whether testosterone rises after a hormone signal. The decision to use it depends on the person's age, earlier results, and specialist practice.

Chromosome testing may be performed, particularly when neither testis is palpable or genital development is atypical. Additional genetic testing may be considered when results suggest a difference in sex development.

Ultrasound and MRI do not always locate abdominal testes and cannot reliably prove that testicular tissue is absent. Imaging should not delay specialist referral. If hormone testing is

uncertain, a urologist may recommend surgery, usually laparoscopy, to inspect the abdomen. Laparoscopy uses a small camera inserted through a small incision. It can identify an undescended testis, ending blood vessels that suggest a testis disappeared before birth, or small remaining tissue.

A newborn with bilateral nonpalpable gonads and atypical genitalia needs prompt specialist evaluation. If the baby also has repeated vomiting, poor feeding, severe sleepiness, or signs of dehydration, emergency care is appropriate. These symptoms can occur with certain adrenal disorders that may initially resemble anorchia.

- Anti-Müllerian hormone, or AMH, which is normally made by supporting cells in the testes
- Inhibin B, another marker of testicular tissue
- Testosterone
- Luteinizing hormone and follicle-stimulating hormone, called LH and FSH, which signal the testes to work

Treatment and management

Testosterone replacement is the main long-term treatment. It replaces a hormone the absent testes cannot make. Treatment is tailored to age, stage of development, health history, and personal goals.

For an adolescent who has not started puberty, testosterone is usually introduced gradually under the care of a pediatric endocrinologist. The goal is to imitate the pace of typical puberty rather than produce all changes at once. Treatment supports growth of the penis, deepening of the voice, muscle development, bone strength, and sexual maturation.

Adults generally need ongoing testosterone replacement to maintain normal hormone effects. Testosterone is available in several forms, including injections, gels, and patches. Each has practical advantages and possible adverse effects. A clinician should explain safe handling, monitoring, and what to do if a dose is missed. Treatment should not be started, stopped, or adjusted without medical guidance.

Monitoring may include testosterone levels, blood counts, growth and pubertal development, blood pressure, and bone health. Adult monitoring may also consider urinary or prostate symptoms when relevant to age and individual risk. Acne, mood changes, skin reactions, or an excessive rise in red blood cells can occur and should be discussed with the prescribing clinician.

If a young child has a very small penis, a specialist may discuss limited hormone treatment

before puberty. This is a separate decision from long-term pubertal treatment and requires individualized assessment.

Testicular prostheses are optional. These implants can create the appearance and feel of testes in the scrotum, but they do not make hormones or sperm. Placement may be considered during childhood, adolescence, or adulthood. Timing depends on body growth, personal preference, and the possibility of future replacement. Potential problems include infection, movement of the implant, discomfort, scarring, or the need for another operation.

Complete bilateral anorchia means sperm cannot be produced. Testosterone therapy does not restore fertility. A reproductive specialist can discuss family-building options, such as donor sperm or adoption, when and if the person wishes.

Psychological support can also be valuable. Concerns about puberty, body image, masculinity, sexual relationships, or fertility are understandable. Counseling should be offered in a respectful way and should not assume that every person experiences the condition in the same way.

Complications

Without adequate testosterone replacement, anorchia can lead to delayed or absent puberty. Over time, low testosterone may contribute to reduced bone density, lower muscle mass, increased body fat, fatigue, sexual difficulties, and reduced quality of life.

Infertility is expected when both testes are completely absent because there is no tissue to make sperm. This may cause grief or stress, especially when the diagnosis or its fertility implications are discussed during adolescence.

Bone health deserves long-term attention. Testosterone helps the skeleton gain strength during puberty and maintain it in adulthood. Delayed treatment, inconsistent treatment, or other risk factors may increase the likelihood of low bone density. Clinicians may recommend a bone-density scan in selected situations rather than for every patient.

Medical procedures can also have complications. Testicular implants may become infected, shift position, or require replacement. Testosterone can cause adverse effects when the dose or formulation is not suitable, which is why regular monitoring is important.

Follow up

Anorchia requires planned, long-term follow-up, usually with an endocrinologist. Children may also see a pediatric urologist, genetics professional, or specialist team experienced in

differences of sex development.

Routine appointments may review:

Seek an earlier routine appointment if puberty is not progressing as expected, treatment side effects appear, doses are often missed, or an implant becomes uncomfortable or changes position.

Urgent medical assessment is appropriate for sudden severe groin or abdominal pain when an undescended testis has not been fully excluded. Emergency care is also appropriate for a newborn with atypical genitalia who develops vomiting, poor feeding, marked weakness, or dehydration. These symptoms may point to another condition that needs immediate treatment.

- Growth and progress through puberty
- Testosterone levels and treatment effects
- Bone health, nutrition, and physical activity
- Blood counts and other treatment-related tests
- Sexual health and fertility questions
- Emotional well-being and body-image concerns
- The condition and position of any prosthetic implants

Sources

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