CONDITION

Imperforate anus

Imperforate anus is a birth difference in which the anus or lower rectum has not formed normally.

WHAT IT IS
Birth difference affecting the anus and lower rectum

BODY SYSTEM
Digestive system, sometimes urinary and reproductive systems

MAIN SYMPTOMS
Missing anal opening, no meconium, swollen abdomen, vomiting

WHAT CAUSES IT
Lower bowel develops differently before birth, usually for unknown reasons

HOW IT IS DIAGNOSED
Newborn physical examination and imaging to map the rectum

MAIN TREATMENT
Surgery, followed by bowel care and long-term follow-up

CAN IT BE CURED?
Sometimes - blockage repaired, bowel-control problems may persist

IS IT AN EMERGENCY?
Sometimes - green vomiting or worsening abdominal swelling

LONG-TERM OUTLOOK
Most children thrive; bowel or urinary care may remain

Overview

Imperforate anus is a condition present at birth. It is part of a wider group called anorectal malformations . These are differences in how the anus and rectum-the last part of the bowel-developed before birth.

The anal opening may be missing, too narrow, or in the wrong place. The rectum may end before reaching the skin. It may also connect to the urinary or reproductive system through an abnormal passage called a fistula .

The exact anatomy differs from child to child. Some malformations are relatively simple to repair. Others involve the bladder, urethra, vagina, spine, or pelvic muscles and need several operations.

The condition matters because stool cannot leave the body through its usual route. A newborn therefore needs prompt assessment by a specialist children's surgical team. Treatment aims to create a safe route for stool while protecting urinary, sexual, and bowel function as the child grows.

Symptoms

Imperforate anus is usually noticed during a newborn examination. Signs may include:

A small opening or narrow anus may be less obvious at birth. It can later cause difficult or painful bowel movements, ongoing constipation, abdominal swelling, or thin stools.

A fistula may allow some stool to leave the body, but this does not mean the bowel has formed normally. The opening may be too small for stool to pass safely.

Some children also have symptoms from related differences in the urinary tract, kidneys, spine, heart, or reproductive organs. These symptoms depend on which other structures are affected.

- No visible anal opening, or an opening in an unusual position
- No passage of meconium, the dark first stool
- Meconium coming from the urethra, vagina, or an opening near the genitals
- Increasing swelling of the abdomen
- Vomiting, which may become green if the bowel is blocked

Causes and risk factors

Imperforate anus develops early in pregnancy when the lower bowel and nearby urinary or reproductive structures do not separate and form in the usual way. In most cases, the exact reason is not known.

It is not caused by anything a parent did or did not do during pregnancy.

Most cases occur without a clear family history. However, anorectal malformations can sometimes be linked to a chromosome or gene change. They may occur as part of a syndrome or alongside a pattern called VACTERL association . This refers to differences involving several areas, which may include the spine, anus, heart, windpipe and food pipe, kidneys, and limbs.

Having a relative with an anorectal malformation may increase the chance of it occurring in a

family, but inheritance is not simple or predictable in most cases. A genetics assessment may be offered when there are several affected relatives, other birth differences, or features suggesting a genetic syndrome.

Diagnosis

A clinician can often identify imperforate anus by examining the newborn's bottom and genital area. The examination looks for an anal opening, its position and size, and any place where meconium is coming out.

Imaging is used to find where the rectum ends and whether there is a fistula. Tests may include an abdominal or pelvic X-ray and ultrasound. If a baby has a colostomy, a contrast study called a distal colostogram may later show the rectum and any fistula in detail before corrective surgery.

More complex malformations may require MRI, endoscopy, or a contrast study of the urinary and reproductive passages. These tests help the surgical team plan treatment rather than simply confirm that the anus is abnormal.

Because related conditions are common, the baby is usually checked for differences elsewhere. This may include:

Finding related differences early can change both the surgical plan and long-term follow-up.

- Kidney and bladder ultrasound
- Spinal examination and imaging
- Heart ultrasound, called an echocardiogram
- Examination of the limbs and food pipe
- Urine and kidney-function tests when needed

Treatment and management

Treatment is planned according to the position of the rectum, the type of fistula, the child's general health, and any other birth differences.

A newborn with a blocked bowel is cared for in hospital. The team may temporarily stop milk feeds, provide fluids through a vein, and use a tube through the nose into the stomach to reduce vomiting and abdominal swelling.

Some simpler malformations can be repaired with one operation during the newborn period.

The surgeon places the rectum within the muscles used for bowel control and creates an anal opening in the correct position.

More complex malformations are often treated in stages. The first operation may create a colostomy , bringing part of the large bowel to an opening in the abdomen. Stool then passes into a bag while the baby grows and the anatomy is studied. A later operation repairs the rectum and anus, after which the colostomy can usually be closed.

A common repair is called a posterior sagittal anorectoplasty , or PSARP. This approach allows the surgeon to see the pelvic muscles and place the rectum within them. Children with a cloaca-a single opening for the rectum, urinary tract, and vagina-need highly specialised assessment and reconstruction.

After healing, caregivers may be taught how to gently dilate the new anus. This helps prevent scar tissue from making it too narrow. Dilations should only be done using the method and schedule provided by the surgical team.

Long-term management may include laxatives, enemas, bowel irrigation, diet changes, or a structured bowel-management programme. These treatments aim for regular emptying and fewer accidents. The right approach is based on the child's anatomy and bowel function rather than age alone.

Complications

Surgery can create a working passage for stool, but it cannot always give normal bowel control. The pelvic muscles, nerves, spine, and sacrum-the triangular bone at the base of the spine-may also have developed differently.

Possible long-term problems include:

A fistula or injury to nearby structures can occasionally cause further surgical problems. Colostomies can also become irritated, narrowed, or prolapsed.

Emotional and social effects may become more important around toilet training, school, and adolescence. Accidents and treatment routines can affect confidence, sleepovers, travel, and relationships. Practical support and privacy are important parts of care.

- Constipation or stool collecting in an enlarged rectum
- Stool leakage or difficulty recognising the need to go
- Narrowing of the repaired anus
- Rectal prolapse, where bowel tissue pushes through the opening

- Skin irritation around the anus or a stoma
- Urinary tract infections or difficulty emptying the bladder
- Kidney problems when major urinary abnormalities are present

Prevention

Imperforate anus usually cannot be prevented. It results from development before birth, and in most cases there is no known action or exposure that caused it.

If the condition appears to be part of a genetic syndrome, or more than one family member is affected, genetic counselling can explain what is known about future pregnancy risks. It cannot always provide an exact estimate.

Prenatal ultrasound may occasionally suggest an anorectal malformation or related birth differences, but many cases are not visible before birth. Screening does not prevent the condition.

Living with it and what to expect

During infancy, care often focuses on surgery, wound healing, anal dilations, and-when present-colostomy care. Follow-up checks growth, stool passage, urinary health, and the function of the repair.

Bowel function becomes clearer over the next several years. Toilet-training age can reveal whether a child senses stool, can hold it, and empties the rectum fully. Early constipation treatment matters because retained stool can stretch the rectum and worsen leakage.

Long-term bowel control is influenced by the type of malformation, the strength of the pelvic muscles, and how well the sacrum and spinal nerves developed. A technically successful repair does not guarantee full continence. Equally, early difficulties do not always show what bowel function will be like later.

Many children attend school, play sports, travel, and take part in everyday activities. Some need only constipation treatment. Others continue a daily bowel programme to stay clean between planned bowel movements. The routine may need adjustment during growth, illness, dietary changes, or puberty.

Children with urinary or reproductive differences may need ongoing urology or gynaecology care. People with cloacal malformations may require monitoring of bladder function, menstruation, sexual health, and future fertility.

As a child becomes a teenager or adult, care should gradually shift to services familiar with adult bowel, urinary, reproductive, and sexual health. Adults born with imperforate anus may still benefit from specialist review, even if childhood surgery was completed years earlier.

When to get medical help

A newborn with no visible anal opening, no passage of meconium, or stool coming from the urinary or genital area needs urgent medical assessment the same day. Do not insert a thermometer, suppository, or enema into an absent or abnormal opening.

Go to an emergency department or call emergency services if a baby or child has:

After surgery, seek urgent advice for fever with worsening wound redness, pus, heavy bleeding, persistent vomiting, marked abdominal swelling, or no stool output when stool would normally be expected. A dark, pale, or suddenly swollen stoma also needs urgent assessment.

Routine follow-up is appropriate for constipation, occasional soiling, skin irritation, concerns about toilet training, or a bowel programme that is becoming less effective. Urinary pain, frequent urinary infections, new leakage, or difficulty emptying the bladder should also be discussed promptly with the specialist team.

- Green vomit
- Rapidly increasing abdominal swelling
- Severe breathing difficulty, unusual sleepiness, or poor responsiveness
- Repeated vomiting with inability to pass stool
- Severe abdominal pain or signs of being very unwell

Common questions

Is imperforate anus the same as anorectal malformation?

Imperforate anus is a commonly used name within the broader group of anorectal malformations. The broader term includes a missing anus, an opening in the wrong place, narrowing, and abnormal connections to urinary or reproductive structures.

Can a baby poop with imperforate anus?

Some babies cannot pass stool at all. Others pass meconium through a small skin opening, the urethra, or an opening near the vagina. Passing some stool does not rule out a serious malformation.

Can surgery completely fix it?

Surgery can create or reposition the anal opening and close abnormal connections. However, bowel control also relies on muscles, nerves, and the spine. Constipation, leakage, or the need for a bowel programme may continue after a successful repair.

Will my child be able to use the toilet normally?

Some children develop voluntary bowel control, while others need medicines, enemas, irrigation, or scheduled bowel care. It may take several years to understand a child's likely level of control. The type of malformation and development of the spine and pelvic muscles are important.

Does imperforate anus affect urination or fertility?

It can. The rectum may connect to the urinary or reproductive tract, and some children have related kidney, bladder, genital, or spinal differences. Effects on sexual function and fertility are more likely with complex malformations, especially cloaca, but they are not the same for everyone.

Can imperforate anus happen again in another pregnancy?

Most cases are isolated, and the exact cause is unknown. The chance may be higher when there is a family history or a known genetic condition. A genetics professional can review the individual family history and available test results.

Sources

- National Institute of Diabetes and Digestive and Kidney Diseases. Anorectal Malformations . U.S. Department of Health and Human Services
- MedlinePlus Medical Encyclopedia. Imperforate anus . U.S. National Library of Medicine
- American Pediatric Surgical Association. Anorectal Malformation . Pediatric Surgery Library
- Wood RJ, Levitt MA. "Anorectal Malformations." Clinics in Colon and Rectal Surgery . 2018;31(2):61-70
- Kyrklund K, et al. "European consensus on the management of patients with anorectal malformations. Part 1: Diagnostics, neonatal management and follow-up." Techniques in Coloproctology . 2020;24:1091-1104

- Holschneider A, et al. "Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations." Journal of Pediatric Surgery . 2005;40(10):1521-1526