

Anal Atresia

Patient Information Series – What you should know, what you should ask.

What is anal atresia?

Anal atresia, also known as imperforate anus, is a congenital abnormality in which there is no opening at the end of the digestive tract, where the anus normally is. It is part of a wider spectrum of abnormalities involving the anus and last part of the bowel, the rectum. Anorectal malformations (ARMs) can range from mild abnormalities with only a thin membrane covering the anus to more severe abnormalities with the rectum ending high up in the belly. The rectum may be connected to the skin or other parts of the body, such as the bladder or the vagina, through a channel, called a fistula.

How does anal atresia happen?

Anal atresia is rare and happens in about 1 in 1,500 to 5,000 live born babies. It is not completely clear how it happens. Approximately half of the babies with an anal atresia will have other problems, most often with their urinary or genital organs, the bones that make up the spine, or heart. Some babies will have multiple problems and have a condition called VACTERL association. This means they have at least three of the following: vertebral abnormalities (the bones of the spine), anal atresia, heart problems, tracheoesophageal fistula (a connection between the windpipe and the feeding tube), kidney or bladder problems and limb abnormalities. These children usually have a normal development and intelligence.

Some babies will have a change in the number of chromosomes or a change in the information within the chromosomes themselves. As an example, people with Down Syndrome have an extra chromosome, number 21. People with Townes-Brocks syndrome have a change in the information in one of the chromosome number 16. Both of those conditions are seen in babies with anal atresia.

Should I have more tests done?

Many women will choose to have more tests done to know more about the condition of the baby. The tests available depend on where you are. Tests to ask about include an amniocentesis to look for problems with the number of chromosomes and some of the problems within the chromosomes. You should also ask if a fetal echocardiography, a specialised ultrasound of the heart of the baby during the pregnancy, can be done.

What are the things to watch for during the pregnancy?

If a baby with anal atresia also has a tracheoesophageal fistula women will accumulate extra amniotic fluid or water around the baby. This condition is called polyhydramnios. It can stretch the uterus too much and cause early labour well before the due date. Knowing this in advance, your doctor or midwife can help with decreasing the risk of an early birth.

What does it mean for my baby after it is born?

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When a baby is born with an imperforate anus, there will be no passage of stool. If there is a fistula, stool will pass through another opening. The doctor will examine the baby to make the diagnosis and determine the severity of the abnormality. Sometimes an ultrasound or X-ray of the belly will be made. Tests to check for problems with the spine, heart and kidneys will be done.

Babies with an anorectal malformation, i.e. cases where anus or rectum has not developed properly, need surgery. The type and number of surgery depends on the type of abnormality your baby has. If the anus is covered by a thin membrane, this is less complex than when the rectum ends higher and connects with the bladder or other structures. The final goal of the surgery is to create an anus in the normal location, repair connections between the rectum and other structures and connect the rectum to the new anus. If the baby needs to grow a bit more before a complex surgery can be performed, a temporary colostomy can be necessary. This is a surgery in which the large intestine is cut and each site of the intestine is diverted through an incision in the abdominal wall. From one opening, stool passes to a pouch that is attached on the outside of the belly, while the other opening lets out normal mucus that is produced by the intestine.

The baby will have to stay in the hospital after surgery. The duration depends on the complexity of the surgery. To prevent the new anus from getting too narrow as it heals, anorectal dilation has to be performed for several weeks. A very small rod-like dilator is used for this purpose and you should be taught how to use this at home.

Babies who also have a problem within the chromosomes may have even more problems after being born. The prognosis and treatment depends on the type of problem that the baby has.

When they grow up, some children develop good bowel control, but the majority will have some bowel problems, such as constipation or fecal incontinence. Follow-up and care by a specialised team, including urologists, gynecologists and gastroenterologists is therefore advised.

Will it happen again?

When no other genetic reason is found to explain the anal atresia, the risk of this happening again is estimated to be approximately 1 in a 100. If there is a genetic reason, then the risk depends on the reason and a consultation with a specialist may be helpful to help sorting this out.

What other questions should I ask?

- What type of anorectal malformation does this look like?
- Do you suspect any other abnormalities?
- Are there abnormalities that are impossible or very difficult to detect before the baby is born?
- How often will I have ultrasound examinations done?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet in advance the team of doctors that will be looking after my baby when it is born?
- What kind of surgery might be indicated?

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