

Cloacal Exstrophy

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

What is cloacal exstrophy?

Cloacal exstrophy is a rare and serious disorder that results in abnormal formation of the abdomen. Fetuses with cloacal exstrophy have an abnormal bladder, bowel present outside of the abdomen, and ambiguous genitalia. Many have spinal, kidney, and limb defects. Cloacal exstrophy is sometimes referred to as the Omphalocele, Exstrophy, Imperforate Anus and Spinal defect complex (OEIS).

How is cloacal exstrophy detected?

Cloacal exstrophy is typically noted during the anatomy ultrasound at 18-22 weeks of gestation but may be detected as early as 12-14 weeks at the time of the nuchal translucency ultrasound.



Cloacal exstrophy seen on ultrasound

What causes cloacal exstrophy?

The exact cause of cloacal exstrophy is unknown, but it is mostly likely caused by abnormal development of the bladder and intestines very early in pregnancy.

Should I have more tests done?

In addition to seeing specialists, you may be offered additional imaging and genetic testing. The most common tests offered include:

- **Detailed ultrasound examination:** This is to carefully look at your baby for any other ultrasound findings or abnormalities. Ultrasound can identify many but not all abnormalities.
- **Fetal echocardiogram:** This is an ultrasound that focuses on the baby's heart. Heart anomalies can be seen in babies with cloacal exstrophy, and knowing if they are present can help guide the care once baby is born.
- **Amniocentesis:** This is a test that removes a small amount of fluid from around the baby by a thin needle. The fluid is tested, and genetic conditions can be diagnosed or confirmed.
- **Cell-free fetal DNA:** This is a maternal blood test that tests your baby's cells from your bloodstream. It is a very good genetic screening test for certain conditions, but it is not as accurate as an amniocentesis. In cases when an amniocentesis is not performed, this could test for some genetic conditions and the sex of the baby.
- **Fetal MRI:** Though ultrasound can detect many anomalies, an MRI can sometimes provide a more detailed look at certain organs.

What should I expect once my baby is born?

Though survival is high, babies with cloacal exstrophy often need multiple surgeries to correct the defects and to attain bowel and bladder function and continence. Many face difficulties with sexual function and bladder and bowel continence. Most babies will require extensive medical care throughout their lives. The prognosis depends on the specific abnormalities found and if there are other abnormalities present.

What other questions should I ask?

- Are there any other abnormalities on the ultrasound?
- What kind of genetic testing will I need to have?
- How often will I have ultrasound examinations done?
- Will my baby need surgery after delivery?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born?