

# Bladder Exstrophy

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

## What is bladder exstrophy?

Bladder exstrophy is a condition that begins during fetal development when the bladder and lower abdominal wall do not correctly form. This causes the bladder to be outside of the baby's body. Bladder exstrophy occurs in 1:30,000-1:50,000 of all births. Bladder exstrophy is often the only abnormality in the pregnancy but sometimes occurs with other conditions.

## How is bladder exstrophy detected?

Bladder exstrophy is often diagnosed by prenatal ultrasound, usually around 20 weeks of pregnancy at the time of the anatomy ultrasound. In fetuses with bladder exstrophy, the bladder is not seen on ultrasound while the kidneys appear normal.



Bladder exstrophy on ultrasound. Image courtesy of Pablo Garcia Manau, MD.

## What causes bladder exstrophy?

In most babies that have bladder exstrophy we are unable to determine why it occurred.

### *Genetics*

Sometimes abnormal changes in the genetic makeup of the baby will cause a condition to occur. We have not found a specific genetic change that occurs in all babies with bladder exstrophy. Most studies suggest that males have a higher risk for bladder exstrophy than females.

### *Environment*

Sometimes various factors in the environment during pregnancy will cause a condition to occur in the baby. We currently do not know if this happens in bladder exstrophy. Some studies have suggested smoking, alcohol, infections, and various medications used in pregnancy may make a baby more likely to have bladder exstrophy. However, there is not enough information at this time to know this for sure.

## Should I have more tests done?

If your doctor is concerned your baby may have bladder exstrophy based on your ultrasound, you will likely be offered additional testing to help confirm the diagnosis and your baby's sex. The exact testing offered will be based on the presence or absence of other ultrasound findings, your medical and pregnancy history, and results from any earlier testing you may have had. You may be referred to a large medical center with a team including a high-risk pregnancy specialist, a pediatric urologist, and a neonatal intensive care unit (NICU) specialist. You may also be offered a consultation with a Genetic Counselor, a medical professional with special training in genetic conditions.

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Tests that may be offered include:

- **A 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.
- **Amniocentesis:** This is a test that removes a small amount of amniotic fluid from around the baby with a thin needle. This can be used to confirm the baby's sex and to perform genetic diagnostic tests.
- **Cell-free fetal DNA:** This is a maternal blood test that uses your baby's cells that are in your bloodstream. It is a very good genetic screening test for certain conditions, such as Down syndrome, but it is not as accurate as an amniocentesis. This test can also determine the sex of the baby.
- **Magnetic resonance imaging (MRI):** A MRI may be suggested to evaluate your baby's bladder, bowel, spine, and genitalia for abnormalities.

## **Bladder exstrophy is often the only abnormality in the pregnancy but sometimes occurs with other conditions. What are these conditions?**

In addition to bladder exstrophy, some babies may have other abnormalities of the abdominal wall, anus, or spine. The intestines, liver, or other organs may also stick outside of the body covered by a thin sac called an omphalocele. The anus may not have an opening to the outside of the body. The baby's kidneys may be affected. In male babies, the testes may not descend, or there may be a problem with the location of the urethra (opening for urine) called epispadias. In females, there may be problems with the structure of the uterus and vagina.

## **If my baby has bladder exstrophy, what will happen after my delivery?**

If your baby is born with bladder exstrophy, he or she will likely need several reconstructive surgeries after birth with a team of specialists. The goal of these surgeries is to place your baby's bladder inside the body and improve the function of your baby's bladder, kidneys, urination, and later sexual function. The baby will require very complex care for many weeks in a specialized hospital.

## **If my baby has bladder exstrophy, will this happen again in my next pregnancy?**

Most cases of bladder exstrophy occur at random, and it is unlikely that your future pregnancies will also be complicated by bladder exstrophy. Most studies report that there is an approximately 1 in 100 chance that this will happen in your next pregnancy.

### **What other questions should I ask?**

- Are there any other abnormalities on the ultrasound?
- What kind of genetic testing will I need to have?
- Do I need additional imaging, such as an MRI?
- 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, prior to delivery?

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