

# Fetal megacystis

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

## **What is a fetal megacystis?**

Fetal megacystis refers to an unusually large urinary bladder seen on ultrasound. This occurs in the antenatal period in about 1 in 1500 pregnancies, much more frequently in boys than in girls. In a first trimester scan (11-14 weeks), megacystis is diagnosed if the longitudinal bladder diameter is more than 7 mm. During fetal life, the fetal kidneys produce urine, which is passed to the bladder through tubes called ureters. The urine is stored in the bladder until it is released through another tube called the urethra, which carries the urine out of the body. In most cases, megacystis results when some condition prevents normal passing of urine.

## **How does a fetal megacystis happen?**

Megacystis is most often caused by obstructive conditions that block the urethra, such as posterior urethral valves, or malformation to the urethra such as stenosis (narrowing or stiffening) or atresia (failure to develop); or persistent cloaca (a complex anatomic developmental malformation). In other cases megacystis is caused by neurologic and/or genetic disorders (i.e. megacystis microcolon intestinal hypoperistalsis syndrome) that prevent the bladder normally passing urine.

When megacystis is seen on fetal ultrasound, it raises suspicion that there may be obstruction (blockage) or constriction (narrowing) of the urethra, which is preventing the urine from leaving the bladder. This is the cause in just over half of cases. Over time, this can lead to there being too little amniotic fluid, or oligohydramnios. The back-up of urine in the bladder can also damage the ureters and kidneys. The amniotic fluid that surrounds and cushions the fetus in the womb is made up largely of fetal urine. As the fetus grows he/she draws the fluid into the lungs to help them develop. The cushioning effect of the fluid around the baby lets his/her muscles and bones develop properly.

In cases of complete obstruction, the inability of urine to be released into the amniotic fluid space around the fetus causes the bladder to enlarge and the amniotic fluid levels to decrease (oligohydramnios), which can result in underdevelopment of the lungs (pulmonary hypoplasia), a life-threatening condition.

There is also a risk of an associated chromosomal abnormalities or specific genetic defects. The prognosis depends upon the underlying cause and severity of the obstruction as well as the presence or absence of other findings.

## **Should I have more tests done?**

If your doctor thinks your baby has a problem that is caused by chromosomal or specific genetic defects, you may be referred for genetic testing and counseling.

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Your doctor will take a sample of cells from the placenta (placenta biopsy, CVS) in the first trimester or a sample of cells from the amniotic fluid (amniocentesis) in the second trimester, to get body tissue, which can be checked for a chromosomal abnormalities or specific genetic defects.

In addition, your doctor may refer you to specialists. You may need more ultrasound scans during the pregnancy.

## **What are the things to watch for during the pregnancy?**

The megacystis detected early can spontaneously resolve or progress over the course of pregnancy. Your caregiver will probably order serial ultrasound scans, to watch carefully to assess whether the megacystis is affecting your baby's growth and well-being. Your caregivers will advise you as the pregnancy progresses.

## **What does it mean for my baby before it is born?**

Management will depend on the underlying cause of the megacystis. A fetal medicine specialist will discuss with you the optimal management suitable to your baby. If the fetus is chromosomally abnormal, there is severe oligohydramnios, the diagnosis is made early in pregnancy or if there is evidence of renal dysplasia on ultrasound, some people decide to terminate (stop) a pregnancy. This is a very personal decision. A healthcare professional will speak with you and support whatever decision you make. For chromosomally normal fetuses, intrauterine prenatal treatment may be feasible.

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

After your baby is born, pediatric urologists and nephrologists (kidney specialists) will examine the baby and consult with you to work out the optimal management suitable to your baby. After thorough evaluation of your child's condition, the team will counsel your family on what to expect in both the short- and long-term.

Postnatal treatment options depend on the type of obstruction. Children with megacystis are more susceptible to kidney infections and kidney failure. Some may eventually require kidney transplant. Some may also experience long-term respiratory symptoms as well as bladder dysfunction, poor growth and musculoskeletal problems. Long-term follow-up is overseen by a pediatric urologist and nephrologist.

## **Will it happen again?**

This depends whether or not a genetic cause was found to explain the megacystis, and what other anatomic findings were present, if any. All these factors affect the likelihood of having

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another baby with megacystis. Your caregiver will likely order early targeted fetal ultrasound examination to rule out the condition in your subsequent pregnancies.

## What other questions should I ask?

- Is it severe?
- How often shall I have ultrasound examinations done?
- Should I have genetic counseling? What genetic testing should we consider?
- 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 care my baby when it is born, in advance of my delivery?

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