

Congenital Megalourethra

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

What is Congenital Megalourethra?

Congenital Megalourethra (CMU) happens when the spongy tissues that run through the shaft of the penis, called "corpus spongiosum" and "corpus cavernosum", giving its shape and erectile function, are partially or entirely absent. The urethra runs below these tissues. Without tissue support, the urethra will dilate and lead to a very large urethra, therefore the name "megalourethra" (from the Greek word "megalo"=large). The urine may accumulate in the megalourethra, making the penis look like a balloon full of urine. As the tissues are lax, passing urine does not happen well. Urine can accumulate in the penis and flow backwards, leading to enlargement of the bladder, the ureters and the kidneys.

If the baby does not pass urine well, eventually, there will be a lack of fluid in the amniotic sac and around the baby. A lack of amniotic fluid can affect the growth of the baby's lungs and gut during the nine months of pregnancy.

How does CMU happen?

It is not clear why CMU occurs. It is a very rare condition with only a few reported cases. More than half of the babies with CMU will have other problems, most often with their heart, gut, or limbs. As several organs can be involved, a thorough anatomical investigation of the fetus should be performed. Depending on the sonographic findings, genetic analyses may follow.

How are chromosomes relevant to CMU?

Chromosomes are the intracellular particles where most of our genetic information is kept. We usually have 46 of them matched in 23 pairs; 23 chromosomes come from each parent. Unique chromosome problems are rarely seen in babies with CMU. However, random genetic errors may be found when chromosomal analysis is performed.

Should I have more tests done?

Many women will choose to have more tests done to know more about their baby's condition. These tests include:

- **Fetal echocardiography:** A specialized sonographic evaluation of the baby's heart during pregnancy.
- **Amniocentesis:** An ultrasound guided invasive procedure to check for problems with the chromosomes (includes the gross analysis called "karyotype" and the detailed analysis called "microarray"). This is done by inserting a fine needle through the abdomen of the mother in the gestational sac and removing a small amount of amniotic fluid surrounding the fetus.

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- **Fetal bladder tap:** An ultrasound guided invasive procedure in which a very fine needle is placed through the baby's belly into the baby's bladder to draw a small amount of urine. A lab will analyze the chromosomes in the fetal cells that are floating in the urine and will also analyze the fetal urine to determine baby's kidney function. Based on the results, a better decision for the mother and the baby can be made.
- **MRI scan:** A scan under strong magnetic fields to create clear images of the baby. MRI is helpful when ultrasound cannot confidently establish the diagnosis, especially in cases when the fluid around the baby is significantly reduced or the baby's position prevents clear imaging.

What are the things to watch for during pregnancy?

Babies with CMU are at risk of problems during pregnancy, and most specialists will recommend regular ultrasound examinations at least every 4 weeks, in order to closely monitor amniotic fluid levels, degree of enlargement of the penis, bladder, ureters and kidneys, and thus indirectly observe the kidneys' function.

Many women will have a reduced amount or even no amniotic fluid around the baby. This condition is called oligohydramnios or anhydramnios. This can lead to lung problems (mainly immaturity) at the time of delivery. Sometimes, in cases where amniotic fluid volume is absent due to obstruction, doctors may decide to insert a drain in the baby's bladder to lead the fluid out, allowing it to re-accumulate around the baby's body. This procedure carries a few risks and is only available in specialized centers.

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

When the baby is born, a large, dilated penis with excess skin is noted. Sometimes, the abdomen is quite enlarged as well, depending on the amount of urine accumulated in the urinary system. Depending on the presence of anhydramnios during pregnancy, and gestational age at delivery, some degree of respiratory discomfort should be expected.

The baby will be assisted by the neonatologist for a full assessment. New blood tests should be done to check the kidneys' function. Once the baby is stable, the pediatric surgeons will assess the baby and discuss with the parents on surgeries necessary to correct the function and cosmetic aspects of the problem. The baby may require admission to an intensive care unit in a specialized hospital and stay admitted for weeks after birth.

When they grow up, babies with CMU may have kidney injury. Depending on the degree of the disease, they may need dialysis to help with kidney filtration, and eventually, a kidney transplant. In addition, when they become adults, many of these individuals may have sexual dysfunction and fertility issues.

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Will it happen again?

If no other genetic reason is found to explain the CMU, the risk of recurrence is minimal.

What other questions should I ask?

- Can I meet the team of doctors that will be assisting my baby when it is born during prenatal care?
- What happens if the kidneys do not function well?
- What happens if a genetic condition is found?
- What problems should I expect as my child grows?
- What kind of surgery will be needed and what result should I expect?

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