# Fetal Aqueductal Stenosis

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

## What is fetal Aqueductal Stenosis?

The fetal brain is surrounded by fluid that circulates around the brain and fills two lateral reservoirs, called ventricles, plus a third ventricle and a fourth ventricle.

Fetal Aqueductal Stenosis (AS) occurs when this normal fluid cannot circulate properly as a result of a blockage in the passage which takes fluid between the third and fourth ventricles.

The blockage causes the two lateral ventricles and the third ventricle to fill with fluid. Although the blockage in the passage is not able to be seen with ultrasound, the extra fluid in the ventricles is easily seen.

## How does AS happen?

Aqueductal stenosis is a rare condition and happens in 1 of every 5000 babies. In most cases, an AS occurs by chance and is not related to any other factors. Some babies with AS are found to have viral infections. Others have genetic causes. Babies with AS can also have other brain problems. 10% of boys with AS have a genetic problem.

## How are chromosomes relevant to an AS?

Chromosomes are where most of our genetic information is kept in our cells. Girls have 46 chromosomes including two X chromosomes, and boys have 46 chromosomes with one X and one Y chromosome. Changes to any of these chromosomes can cause problems in the baby as it is developing.

When a boy fetus has a genetic problem causing AS, it is usually linked to the sole X chromosome. Since boys only have one X chromosome, if there is a problem in the X chromosome, it is more likely to cause problems than if a girl has a problem in the X chromosome. About 10% (1 in 10) boys with AS have an X chromosome problem. A clue that a baby might have an X chromosome problem is an abnormal position of the baby's thumbs, but this is seen in only half of boys with an X chromosome problem.

**Should I have more tests done?**

You will be offered additional testing to help determine the reason for the aqueductal stenosis. 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 also be offered consultation with a Genetic Counselor, a medical professional with special training in genetic conditions.

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 fluid from around the baby by a thin needle. The fluid can be tested for chromosomal abnormalities as well as fetal infections.
- **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.
- **Maternal blood tests for viral infections** such as cytomegalovirus or toxoplasmosis: These tests may determine if you have had a recent or past infection, but they will not tell you if your baby has been infected. If your results show a possible infection, additional testing may be recommended to confirm an infection in your baby.
- An **MRI scan** may also be suggested to get a closer look at the baby's brain. This scan uses magnetic fields and radio waves to create pictures of the body. It does not have radiation and is considered safe in pregnancy. However, a fetal MRI may not be available in the area where you live, and it may not change the management of the pregnancy even if it is done.

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

- *Ventriculomegaly/Hydrocephalus:* When AS is present the backup of fluid may get worse during the pregnancy. The areas with extra fluid, the ventricles, can continue to get bigger and push on the surrounding brain. When the ventricles are large, it is known as **ventriculomegaly**.

  Sometimes, the whole head of the baby gets large. This is known as **hydrocephalus**. If the baby's head gets too big to fit through the birth canal, a caesarean delivery may be suggested or a delivery earlier than 39 weeks.

- *Stillbirth:* Babies with AS have a 1-3 in 10 chance of having sudden death during pregnancy. Monitoring the pregnancy in a specialised center is recommended but these events may not be preventable.

For these reasons, extra ultrasounds during pregnancy to watch baby's growth and wellbeing may be necessary.

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

Babies with AS should be born in a hospital where the baby can have specialized care after delivery.

After delivery, the baby will have additional pictures taken of the brain. A neurosurgeon may be consulted to determine if surgery is needed. These procedures are often done within the first few weeks of life to help drain the extra fluid.

Many babies with AS have normal developmental outcomes. Others have delays in development and learn slower than other children. About half of the babies have seizures (a diagnosis known as epilepsy) and need medications to manage seizures. There is no test that can predict a normal outcome.

It will be important to take your baby to the pediatrician to watch for developmental delays, seizures, and changes to the baby's eyesight. The pediatricians will help monitor the baby's development. They can also work with the neurologist to make a plan for taking pictures of baby's brain as the baby grows up.

**Will it happen again?**

If this is not related to a problem with the X chromosome, there is about a 4% (4 in 100) chance of AS happening again. If an issue with the X chromosome is found, there is a 50% chance of this happening again in another boy baby. If other genetic problems are found there may be a 25% chance for this to happen again. Consultation with a genetic counselor can be helpful to figure out the risks for your next pregnancy.

In any future pregnancy, a detailed ultrasound in the second trimester and a follow up scan in the third trimester should be performed since some cases of AS are not able to be identified early in the pregnancy.

**What other questions should I ask?**

- Do you see other changes in the baby's brain?
- How big are the ventricles in baby's brain?
- How big is the baby's head?
- Is my baby a boy or a girl?
- Have you been able to see the position of the baby's thumbs?
- 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 the team of doctors that will be assisting my baby when it is born in advance?


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