

What is Oesophageal Atresia?

Oesophageal atresia (OA) is diagnosed when the oesophagus (feeding tube) does not normally connect to the stomach. A tracheoesophageal fistula (TEF), which is an abnormal connection between the trachea and the oesophagus, may also be present.

How is Oesophageal Atresia?

It is not clear why some fetuses develop oesophageal atresia. About half of fetuses with oesophageal atresia are otherwise normal and this is the only problem that the fetus has. The other half of fetuses with OA have other malformations, most often involving their heart or intestines. One out of 10 cases are due to abnormalities in the number of the chromosomes. Common chromosomal abnormalities in fetuses with oesophageal atresia include Down Syndrome (an extra chromosome number 21), Edwards Syndrome (an extra chromosome number 18) and CHARGE syndrome (a change in the information of one of the number 8 chromosomes). OA happens in about 1 of every 4000 babies.

Should I have more tests done?

You will be offered additional testing to evaluate the baby in detail and to make sure that the remainder of the baby appears normal. 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, and a Pediatric Surgeon, to discuss management of your baby after birth.

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.
- **Fetal echocardiogram:** This is detailed evaluation of the baby's heart using ultrasound.
- **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. Other genetic tests may be offered, such as Chromosomal Microarray (CMA, or "chip") which looks more closely at the genetic make-up of the fetus.
- **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.

- **Fetal MRI:** This is a special scan which is sometimes performed to evaluate the baby further.

What are the things to watch for during the pregnancy?

Most specialists recommend regular ultrasound examinations during pregnancy. Ultrasound can help in identifying if the baby has other structural anomalies. Extra amniotic fluid or “water” may also accumulate around the baby. This condition is called polyhydramnios. It can be very uncomfortable, cause early labour and sometimes delivery before the due date. Knowing if there is polyhydramnios through regular ultrasound examinations, your doctor or midwife can help with decreasing the risk of an early birth.

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

After the baby is born, a suction catheter will be placed in the upper part of the oesophagus to drain secretions. This will prevent fluid passing to the lungs through a tracheoesophageal fistula and prevent the baby from inhaling fluid into its lungs. An X-ray or MRI may be performed to confirm the diagnosis and make sure there are no other abnormalities not able to be seen before delivery.

A surgeon will be consulted. Typically, babies with OE have surgery soon after birth. The goal of the surgery is to connect the esophagus to the stomach, and to close the tracheoesophageal fistula, if one is present.

Babies with OE sometimes develop gastroesophageal reflux and lung diseases. Your pediatrician will help identify and treat these problems as the baby grows.

Babies who have chromosomes problems or other malformations may have even more problems after being born. Treatment for these special babies will be individualized.

Will it happen again?

When no other genetic reason is found to explain the OA, the risk of this happening again is less than 1 chance in a 100. If there is a “genetic” explanation for the OA, then the risk depends on the specific cause. Consultation with a genetic specialist may be helpful.

What other questions should I ask?

- Are there any other abnormalities on the ultrasound?
- What kind of genetic testing is recommended?
- How often will I have ultrasound examinations done?
- When is surgery typically done for babies with OA?
- Where should I deliver?
- At what gestational age should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet in advance the team of doctors who will be looking after my baby when it is born?