

Ectopia Cordis

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

What is Ectopia Cordis?

Ectopia cordis (meaning “heart out of place”) is a congenital anatomic defect that occurs when the anterior chest wall does not develop completely. This results in the heart, partially or completely, being outside the fetal chest.

How does Ectopia Cordis happen?

It is not clear why ectopia cordis occurs. It is a rare condition and happens in about 6 in one million babies. In most cases, it occurs randomly. Most babies with ectopia cordis will present with other problems, most often abnormalities in the heart and abdomen. In some cases, there can be changes in the number of chromosomes, or information within the chromosomes themselves.

How are chromosomes relevant to Ectopia Cordis?

Chromosomes are where most of our genetic information is stored. We usually have 46 of them matched in pairs: 23 come from one parent and 23 from the other parent. For example, people with Down syndrome have an extra chromosome 21. Babies with ectopia cordis can present with various changes in this information, such as having an extra chromosome 18, lacking an X chromosome or having an extra complete set (69 chromosomes).

Should I have more tests done?

Many women will choose to have more tests done to know more about the condition of their baby. You may be able to consult with a geneticist, a specialist in genetics and congenital disease, to help you determine the best approach for your fetus and your family. You can consult with your caregiver and/or a maternal-fetal medicine specialist to decide whether terminating the pregnancy is the right choice for you and your family.

The tests and consultations available depend on where you are. Tests to ask about include:

- an **amniocentesis** to look for problems with the number of chromosomes and some of the problems within the chromosomes. This is done by removing a small amount of the amniotic fluid surrounding the fetus. 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, and whole exome sequencing, which provides even more details.
- a **fetal echocardiography** - a specialised ultrasound of the heart of the baby during the pregnancy, which uses sound waves that “echo” off the structures of the fetus' heart.
- If available, an **MRI scan** can sometimes be performed to provide information on the condition of the baby. This scan uses strong magnetic fields and radio waves to create detailed images of internal body structures.

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What are the things to watch for during my pregnancy?

Babies with ectopia cordis are at risk of various problems during pregnancy. Most specialists will recommend regular ultrasound examination, often every 2 weeks. The ultrasound will help identify if the baby is going into heart failure due to the pressure on the heart outside the chest.

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

A baby with the heart outside the chest wall has a high risk of other heart defects that may require surgery after birth, and this is the biggest problem for babies with ectopia cordis. If there are associated heart defects, the baby will probably find it hard to survive. If the heart is anatomically normal, doctors may attempt to introduce it inside the chest, however this is difficult because, in most cases, it will not fit correctly. If the chest is too small to allow the heart to be introduced, the baby will usually die.

Babies who also have had altered information within their chromosomes may have other problems after being born. These outcomes will depend on the specific genetic disorder.

If the baby is stable, a surgeon will perform one or more surgeries to attempt to put the heart back into the chest. If this is possible, the baby will require very complex care in a specialised hospital for weeks after birth.

Will it happen again?

If no other genetic reason is found to explain ectopia cordis, the risk of it happening again is very low. If there is a genetic reason, this will determine the risk, and a consultation with a genetic specialist may be helpful to sort this out.

What other questions should I ask?

- Is this complete or partial ectopia cordis?
- Is the heart normal, or are there other heart defects?
- Are there other defects besides ectopia cordis?
- How often will I have ultrasound examinations done?
- If I want it, is termination of pregnancy an option?
- 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 after birth, before delivery?

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Last updated August 2024