Heterotaxy Syndrome

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

What is Heterotaxy syndrome?

A Heterotaxy Syndrome is diagnosed when there are differences in how the organs in the chest and abdomen are positioned. Certain organs, such as the heart and stomach, are normally located on one side of the body. When these organs are not in their usual position, there may be a concern for a heterotaxy syndrome. Some other terms that may be used in describing a heterotaxy syndrome are left atrial isomerism and right atrial isomerism, or polysplenia and asplenia.

The types of problems or malformations that a baby will have with heterotaxy syndrome are varied but can include problems with how the heart is formed (congenital heart defects), how the organs in the abdomen work, and how the electrical system of the heart works. Some babies with heterotaxy have many major problems, and some have only mild problems. Your doctor will be able to explain what the specific concerns are for your baby.

How does Heterotaxy syndrome happen?

During very early fetal development, the body organizes organs and structures into their normal positions, some on the right of the body and some on the left. The heart also develops with certain parts on the left and other parts on the right. With a heterotaxy syndrome, there has been a very early difference in this arrangement that occurred, usually before 3 weeks of pregnancy have elapsed.

What are the possible causes?

There is no known single cause for most cases of heterotaxy syndrome.
There are some genetic conditions that can be associated with heterotaxy syndrome and some of these can be tested for. Poorly controlled diabetes prior to or in early pregnancy may be associated with heterotaxy syndrome. Some medications or drug exposures may be associated with heterotaxy syndrome, including some narcotics or cocaine use.

Should I have more tests done?

- A high-level ultrasound of the fetus is recommended to look for other structural problems in other organ systems.
- A detailed ultrasound of the fetal heart (a fetal echocardiogram), and consultation with a Pediatric Cardiologist is offered for review of fetal heart structure and counselling.
- Referral to a genetics specialist and/or genetic testing may be recommended.
- A fetal MRI may be offered.

What are the things to watch for during pregnancy?

If a heterotaxy syndrome is suspected, you will probably be monitored by a high-risk obstetrician/Maternal-Fetal Medicine Specialist. You will have close monitoring by ultrasound to assess the growth and well-being of your baby. If there are heart problems in your baby, a Pediatric Cardiologist (heart doctor) will also usually follow your baby regularly during pregnancy.

What should I expect after delivery?

Many babies will need to be delivered at a “high-risk” or tertiary care center, where specialists in Newborn Medicine and Cardiology can see and treat the baby after birth. Most babies will have tests after birth to look at the heart and other organs, including ultrasounds, heart echocardiograms, and x-rays. Some babies with congenital heart defects may need heart surgery. Some babies will go home on a preventative antibiotic medication if there are concerns about their spleen.

Will it happen again?

There is a low chance of a heterotaxy syndrome happening in a future pregnancy. If there has been a genetic cause found on testing, your genetics doctor or counselor can advise you on whether there is a risk in future pregnancies.

How can I prevent this?

If you are diabetic, your blood sugars should be well controlled before planning to get pregnant.

Discuss any medication use prior to or in pregnancy with your health care professional. A consultation with a Maternal-Fetal Medicine Specialist prior to planning the next pregnancy is recommended to review any risk factors.

What other questions should I ask?

- Who are the members of the team who will be looking after my baby after birth?
- Will my baby need surgery? What type and when?
- What can I expect for my baby in the months after birth and as he or she grows up?

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