

Heterotaxy syndrome



Boston Children's Hospital
Cardiology



Introduction



What is heterotaxy syndrome?

Heterotaxy syndrome is a rare condition in which some of the body's organs may be in an abnormal place or formed abnormally, and may not work the right way. The organ systems most commonly affected are the heart, lungs, immune system and the gastrointestinal (GI) tract.

How common is heterotaxy syndrome?



Heterotaxy syndrome happens in about **1 out of every 10,000** live births.

What causes heterotaxy syndrome?

- Heterotaxy syndrome is caused by abnormalities in a child's genes. These abnormalities may be inherited from one or both parents. Or it may happen at random.
- Blood tests can detect a specific gene that may cause heterotaxy in some children. In many others, a genetic cause can't be identified.
- A positive genetic testing result can help understand a child's prognosis (outlook) and risk for complications outside of their heart. It can also be used to understand if other family members are at risk for having the condition and the likelihood that the parents of a child with heterotaxy could have a second affected child.

How is heterotaxy syndrome diagnosed?

Heterotaxy syndrome is diagnosed by identifying characteristic findings in multiple organ systems. In many cases, there's not one specific way of diagnosing heterotaxy. An expert in heterotaxy may need to help identify whether your child has the diagnosis.

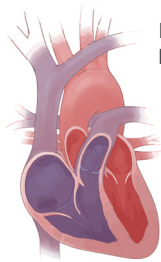
How does it affect the body?



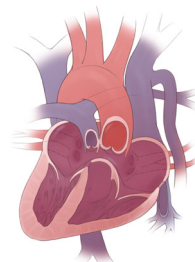
Heart

Heart defects range from mild to severe and can include:

- Abnormal heart position, such as dextrocardia (right-sided heart) or mesocardia (heart in the middle of the chest)
- Abnormal location of major blood vessels that carry blood to and from the heart
- Abnormal development of the heart chambers or abnormal connections between the chambers
- Abnormalities of the electrical system of the heart, causing fast or slow heart rates



Normal heart



Example of heart with heterotaxy syndrome



Lungs and airways

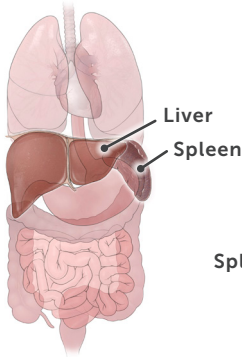
Lung and airway abnormalities can include:

- Abnormally shaped airways or normally shaped airways in the wrong position
- Primary ciliary dyskinesia (PCD), a lung condition in which the cilia, the tiny hair-like cells that line the respiratory tract, are not formed the right way or may not work properly. This can cause chronic nasal or chest congestion, pneumonia and abnormal dilation of the airway called "bronchiectasis"

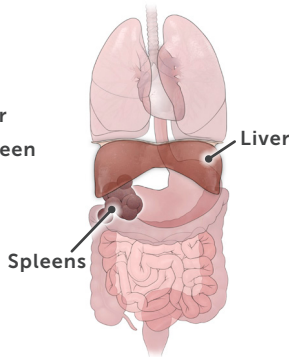


Immune system

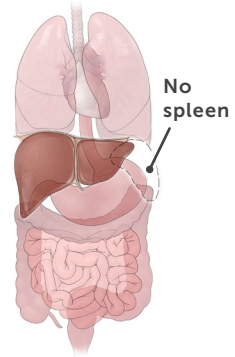
The spleen may not exist, be small or in the wrong place. This can put a child at higher risk for infections.



Normal



Polysplenia



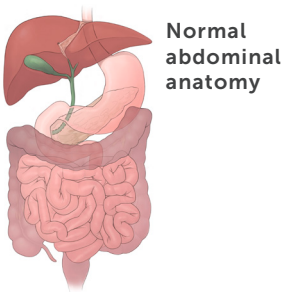
Asplenia



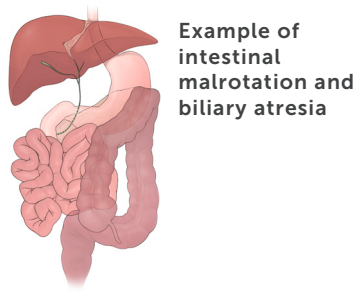
GI tract

GI tract abnormalities can include:

- The liver may be in the wrong place (either on the left side of the abdomen or in the middle)
- The intestines may not have rotated properly before birth, called "malrotation," and may be at risk of abnormally twisting upon itself



Normal abdominal anatomy



Example of intestinal malrotation and biliary atresia

What tests might my child have?

There are many screening tests that help your child's care team establish a diagnosis and better understand how heterotaxy syndrome impacts your child. The care team will talk to you about which tests are best for your child.



Heart

- **Echocardiogram** – an ultrasound of the heart to evaluate the heart's structure and function
- **Cardiac MRI or CT** – a scan of the heart that can provide additional details about the heart's structure and function
- **Electrocardiogram** – a brief snapshot of the electrical activity of the heart
- **Ambulatory heart rhythm monitor** – a continuous recording of the electrical activity of the heart that lasts anywhere from 1 to 14 days



Immune system

- **Abdominal ultrasound** – an ultrasound of the abdomen to see where the abdominal organs are, such as the spleen, liver and stomach
- **Nuclear medicine spleen scan** – a test that sees how well the spleen is working
- **Blood tests** can tell how the immune system and spleen are working. These include a complete blood count (CBC) with differential, a smear to look for Howell-Jolly bodies, immunoglobulin levels, pitted RBC count, antibody titers and lymphocyte assessments.



Lungs

- **Bronchoscopy** – a test in which a small, flexible tube with a camera called a “bronchoscope” is inserted into the airway to evaluate the airway structure and can also sample fluid in the airways. Your child will be under anesthesia for this test.
- **Ciliary biopsy** – a test that collects a small amount of cells from the inside of the nose or the inside of the airways to evaluate the structure of the cilia and screen for PCD
- **Nasal nitric oxide measurement** – a test that measures the amount of nitric oxide in your child’s nose



GI tract

- **Upper GI study** – a test in which your child drinks contrast or contrast is given through a tube directly into your child’s stomach to look for malrotation and/or a volvulus
- **Swallow study** – a test in which your child drinks contrast to evaluate the safety of their swallowing



Genetics

- **Chromosomal microarray** – a blood test that looks at all of your child’s chromosomes (the structures inherited from each parent that contain our genes) to look for missing or extra pieces of genetic material that can cause heterotaxy syndrome
- **Heterotaxy and PCD Gene Sequencing** – a blood test that looks at the individual DNA letter sequence of genes known to cause heterotaxy syndrome and PCD

What special care might my child need?

- The care that your child needs will depend on which body systems are affected and how severely they are affected.
- Some children with heterotaxy syndrome who have severe heart defects may need heart surgery shortly after birth to stabilize their circulation. Other children are only mildly affected and never need any type of surgery.
- Children with PCD may need special inhaled medications to keep their lungs healthy. Children with immune system problems may need daily antibiotic treatment and extra vaccinations to prevent certain infections. Children with malrotation who develop an intestinal blockage may need surgery to correct this. Test results will be reviewed with you by your child's care team and a treatment plan will be developed that's best for your child.

Who is on my child's heterotaxy team?

The team includes a dedicated nurse practitioner and physician cardiology team, as well as physicians from the related specialties, including Pulmonary, Immunology, General Surgery and Genetics.

We understand that this information may seem overwhelming. The Boston Children's Hospital heterotaxy team is here to help you starting from the time when your child is diagnosed. We partner with you to make sure that your child gets the appropriate care and treatment. Children with heterotaxy syndrome often live healthy and productive lives, and it's our goal to make sure that your child has the best chance at having a bright future.

Resources

Where can I get more information?

<https://www.childrenshospital.org/conditions/heterotaxy>

<https://heterotaxyconnection.org>

(support group dedicated to educating and empowering families affected by heterotaxy syndrome)

Contact Us

Email: heterotaxy@childrens.harvard.edu

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