Heterotaxy

A guide to your child's care

Boston Children's Hospital
Cardiology

Introduction

What is heterotaxy?

Heterotaxy 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**, the **lungs**, the **immune system** and the **gastrointestinal system.**

How common is heterotaxy?

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

What causes heterotaxy?

- Heterotaxy is caused by variations in a child's genes. These variations may be inherited from one or both parents, or they may occur for the first time in the affected child.
- Blood tests can detect a specific gene change that may cause heterotaxy in some children. In many others, a genetic cause cannot be identified.
- A positive genetic test result can help understand a child's prognosis (outlook) and risk for complications outside of the heart. It can also be used to understand if other family members are at risk for having heterotaxy and the likelihood that the parents of a child with heterotaxy could have a second child who is affected.

How is heterotaxy diagnosed?

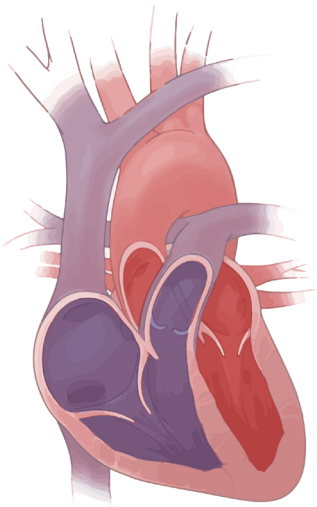
Heterotaxy is diagnosed by identifying characteristic findings in multiple organ systems. Some children are diagnosed before birth, while others are not diagnosed until infancy or childhood. A heterotaxy expert may need to help identify whether your child has the diagnosis.

How does it affect the body?

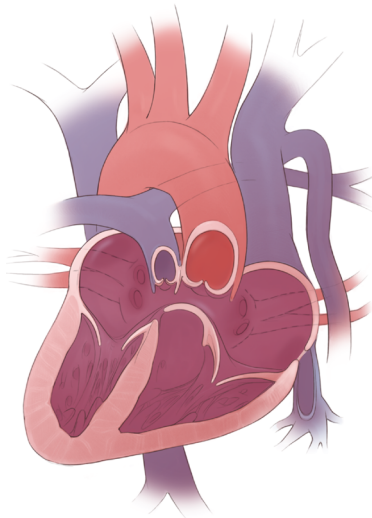
Heart

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



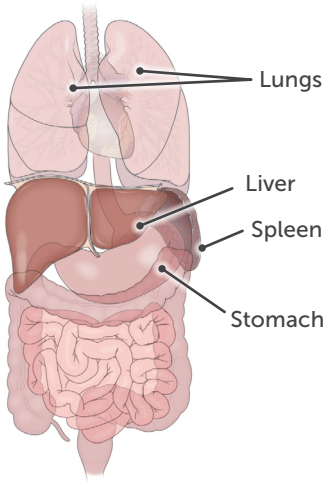
Normal heart



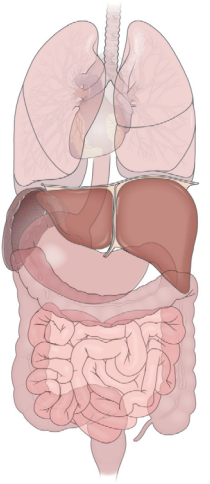
Complex congenital heart defect

Immune system

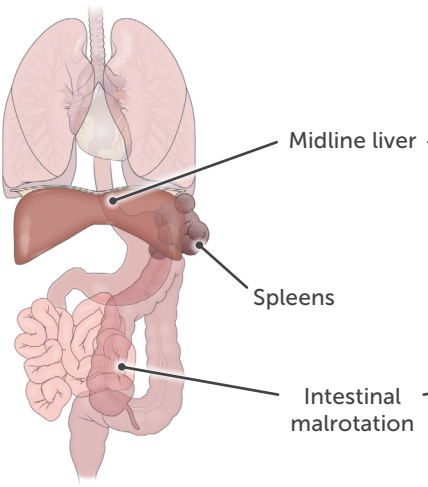
The spleen plays an important role in fighting infections. In heterotaxy, there may be no spleen (*asplenia*) or several small spleens (*polysplenia*). In some cases, the spleen is smaller than usual or in the wrong location. Abnormal spleen function can put a child at higher risk for getting an infection.



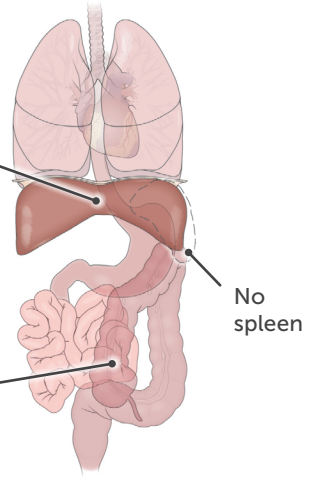
Situs solitus (normal)



Situs inversus (mirror image of normal)



Polysplenia

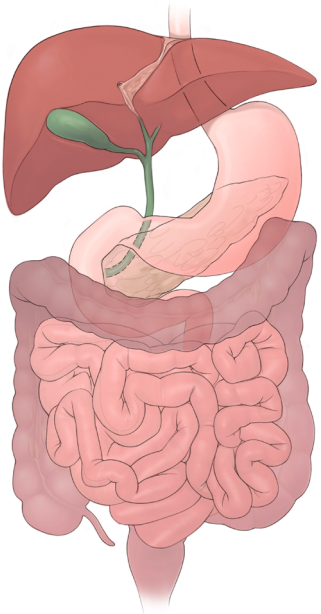


Asplenia

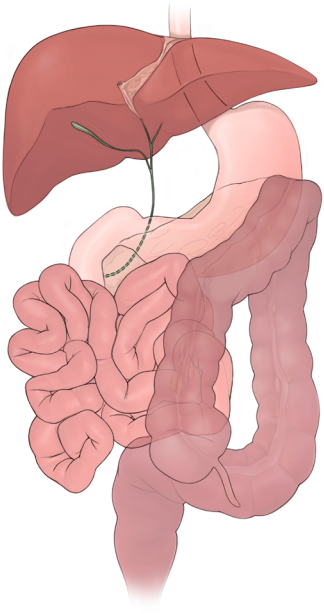
Gastrointestinal (GI) system

Gastrointestinal abnormalities can include:

- The intestines may not have rotated properly before birth, called *malrotation*, and may be at risk of abnormally twisting upon itself and becoming blocked, called *volvulus*.
- The bile ducts may be blocked or underdeveloped, causing bile to build up in the liver. This is called *biliary atresia*.



Normal abdominal anatomy

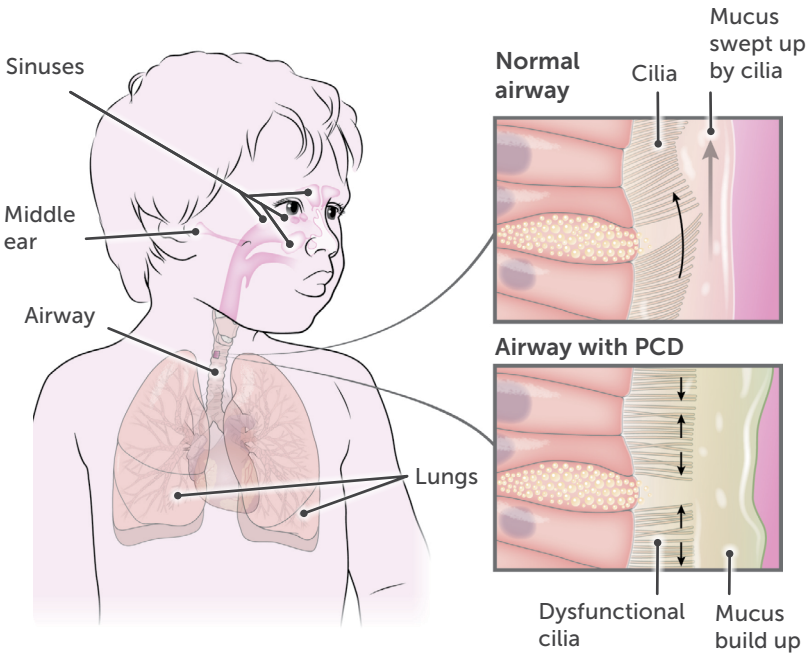


Intestinal malrotation and biliary atresia

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*.



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 impacts your child. The care team will talk to you about which tests are best for your child.

Heart

Electrocardiogram – a brief snapshot of the electrical activity of the 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

Cardiac catheterization – a procedure where catheters (small tubes) are placed inside the heart to check pressures and take detailed pictures

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

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 and airways

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 and sinuses to screen for PCD

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

Gastrointestinal system

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

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

Heterotaxy and PCD Gene Sequencing – a blood test that looks at the individual DNA letter sequence of genes known to cause heterotaxy and PCD

Exome or Genome Sequencing – a blood test that looks at all of the genes for DNA sequence changes and extra or missing pieces of chromosomes, and often includes samples from the parents for comparison

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, gastroenterology, cardiac surgery, general surgery, and genetics.

We understand that this information may feel overwhelming. The Boston Children's Hospital Heterotaxy Program is here to help you from the moment your child is diagnosed. We partner with you to ensure that your child receives the appropriate care and treatment. Children with heterotaxy often lead healthy and productive lives, and our goal is to provide your child with the best chance for a bright future.

Resources

Where can I get more information?

Boston Children's Hospital Heterotaxy Program webpage:
childrenshospital.org/conditions/heterotaxy

Heterotaxy Connection (support group dedicated to educating and empowering families affected by heterotaxy):
heterotaxyconnection.org

Heterotaxy Research and Clinical Care Collaborative / HRC3 (a nationwide collaborative of physicians and scientists working together to better understand and improve the outcomes of children living with heterotaxy):
hrc3.org

Contact us

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Scan QR code to learn more about Boston Children's Heterotaxy Program

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