
Facts about Congenital Heart Defects

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Congenital heart defects are conditions that are present at birth and can affect the structure of a baby's heart and the way it works. They can affect how blood flows through the heart and out to the rest of the body. Congenital heart defects can vary from mild (such as a small hole between the chambers of the heart) to severe (such as missing or poorly formed portions of the heart).

Signs and symptoms for congenital heart defects depend on the type and severity of the particular defect. Some defects might have few or no signs or symptoms, while others might cause a baby to have bluish tinted nails or lips, fast or troubled breathing, to tire easily when feeding, or to be very sleepy.

Congenital heart defects are

- The most common type of birth defect.
- A leading cause of infant death.
- Present among nearly 40,000 births in the United States each year.

Diagnosis

Some congenital heart defects may be diagnosed during pregnancy using a special type of ultrasound called a fetal echocardiogram, which creates pictures of the heart of the fetus. However, some congenital heart defects are not detected until later in life, during childhood or adulthood. Usually, though, congenital heart defects are diagnosed at birth or shortly afterward. If a health care provider suspects a congenital heart defect is present, the baby will have several tests (such as blood tests, an X-ray, and an echocardiogram) to confirm the diagnosis.

In New Jersey, a newborn baby will have a Pulse Oximetry test before he/she is sent home. This simple painless test measures the amount of oxygen in the blood and may alert the doctor to a possible congenital heart defect.

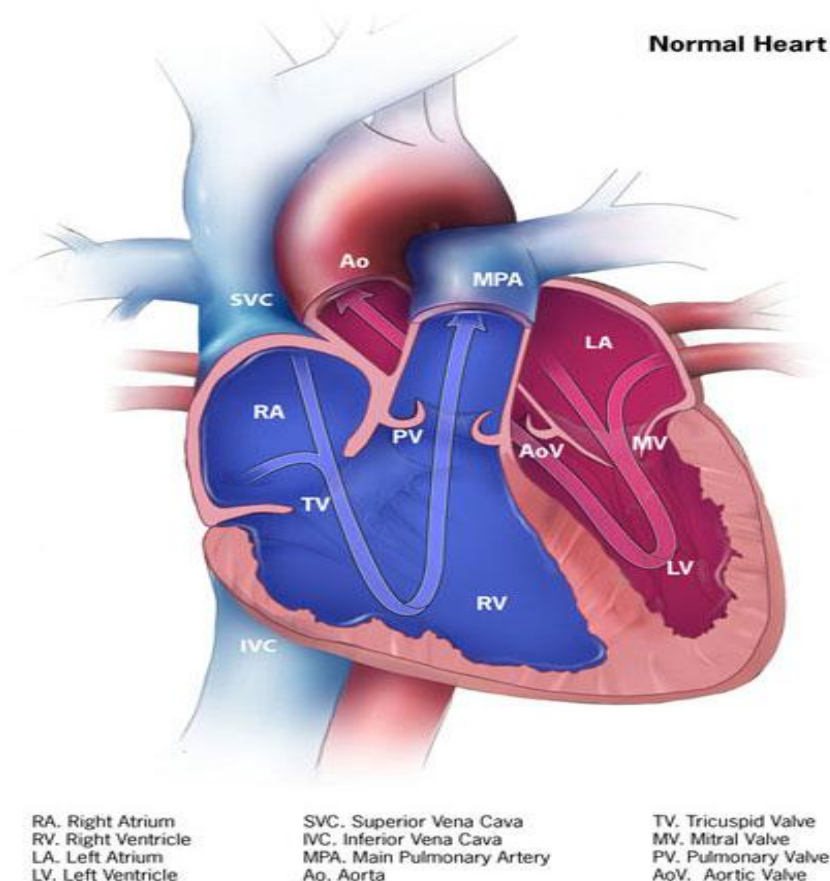
Fetal development

A fetus's heart develops early (3-5 weeks after conception). It starts out as a tube and then grows into 4 chambers around 5 weeks. It starts beating around 5 weeks but usually can't be heard until 7-9 weeks.

The change in the heart from a simple tube to a complex organ takes several steps. In the first step the simple tube forms and grows. During the next step, the tube grows too big for its space and folds and twists to form the basic shape of the heart. As the heart continues to grow, the two top chambers (atria) of the heart form and separate. Finally, in the last step, the bottom chambers (ventricles), which pump the blood, grow and separate into two separate chambers.

Blood circulation in the fetal heart is different than in the heart of the newborn baby. It is different because the fetus's oxygen comes from the mother's blood, while a newborn baby gets oxygen from the air inhaled into his/her lungs. To help send oxygen rich blood from the mother to the developing fetus, there are two openings (patent ductus arteriosus and patent foramen ovale) in the fetal heart that are not present in a newborn baby's heart. Around the time of birth, these openings close so blood can be pumped to the newborn baby's lungs to get oxygen.

Below is an image of a normal heart.



This image was provided by the Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities.

Known Causes

The causes of congenital heart defects among most babies are unknown. Some babies have heart defects because of changes in their genes or chromosomes. Congenital heart defects also are thought to be caused by a combination of genes and other risk factors, such as exposures to substances in the environment, maternal diet, or maternal medication use.

The Center for Disease Control and Prevention (CDC) continues to study congenital heart defects to learn how to prevent them. Many structures in the baby (such as the heart) often are formed before many women realize they are pregnant. CDC's study collaborators have reported important findings about some pregnancy exposures that increase the risk for congenital heart defects: obesity, diabetes, and smoking. If a woman is obese, has diabetes, or smokes and is pregnant or thinking about getting pregnant, she should talk with her doctor about ways to increase her chances of having a healthy baby.

Preventative Actions/Activities

Not all birth defects can be prevented, but studies have shown there are actions/activities a woman can take to increase her chances of having a healthy baby. These actions/activities include:

- Take 400 micrograms (mcg) of folic acid every day for at least 1 month before getting pregnant to help prevent birth defects.
 - Folic acid is also in some fortified foods like cereal.
- Stop smoking and drinking alcohol.
- Do not use “street” drugs.
- If you have a medical condition, be sure it is under control. Some examples of medical conditions include asthma, diabetes, oral health, obesity, and epilepsy. Also be sure that your vaccinations are up to date.
- Talk to a health care professional about any over-the-counter and prescription medicines you are taking. These include dietary or herbal supplements.
- Avoid contact with toxic substances or materials that could cause infection at work and at home. Stay away from chemicals and cat or rodent feces.
- See your health care provider if you are considering becoming pregnant or as soon as you think you may be pregnant
- Eat a healthy diet.

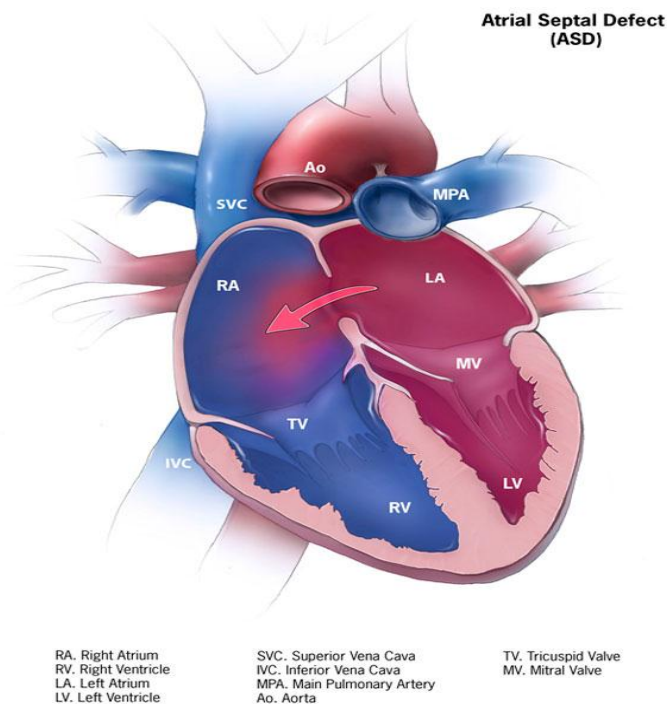
Examples of Specific Congenital Heart Defects

- **Atrial Septal Defect**

An atrial septal defect (ASD) is a birth defect of the heart in which there is a hole in the wall (septum) that divides the upper chambers of the heart (atria). A hole can vary in size and may close on its own or may require surgery. The hole increases the amount of blood that flows through the lungs and over time, it may cause damage to the blood vessels in the lungs. Damage to the blood vessels in the lungs may cause problems in adulthood, such as high blood pressure in the lungs (pulmonary hypertension) and heart failure. Other problems may include abnormal heartbeat, and increased risk of stroke.

Though an atrial septal defect is present at birth, many babies do not have any signs or symptoms. In fact, it is possible that an atrial septal defect might not be diagnosed until adulthood. One of the most common ways an atrial septal defect is found is by detecting a murmur when listening to a person's heart with a stethoscope. If a murmur is heard or other signs or symptoms are present, the health care provider might request one or more tests to confirm the diagnosis. The most common test is an echocardiogram which is an ultrasound of the heart.

Treatment for an atrial septal defect depends on the age of diagnosis, the number of or seriousness of symptoms, size of the hole, and presence of other conditions. Sometimes surgery is needed to repair the hole. Sometimes medications are prescribed to help treat symptoms. There are no known medications that can repair the hole.



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- ***Hypoplastic Left Heart Syndrome***

Hypoplastic left heart syndrome (HLHS) is a birth defect that affects normal blood flow through the heart. As the baby develops during pregnancy, the left side of the heart does not form correctly.

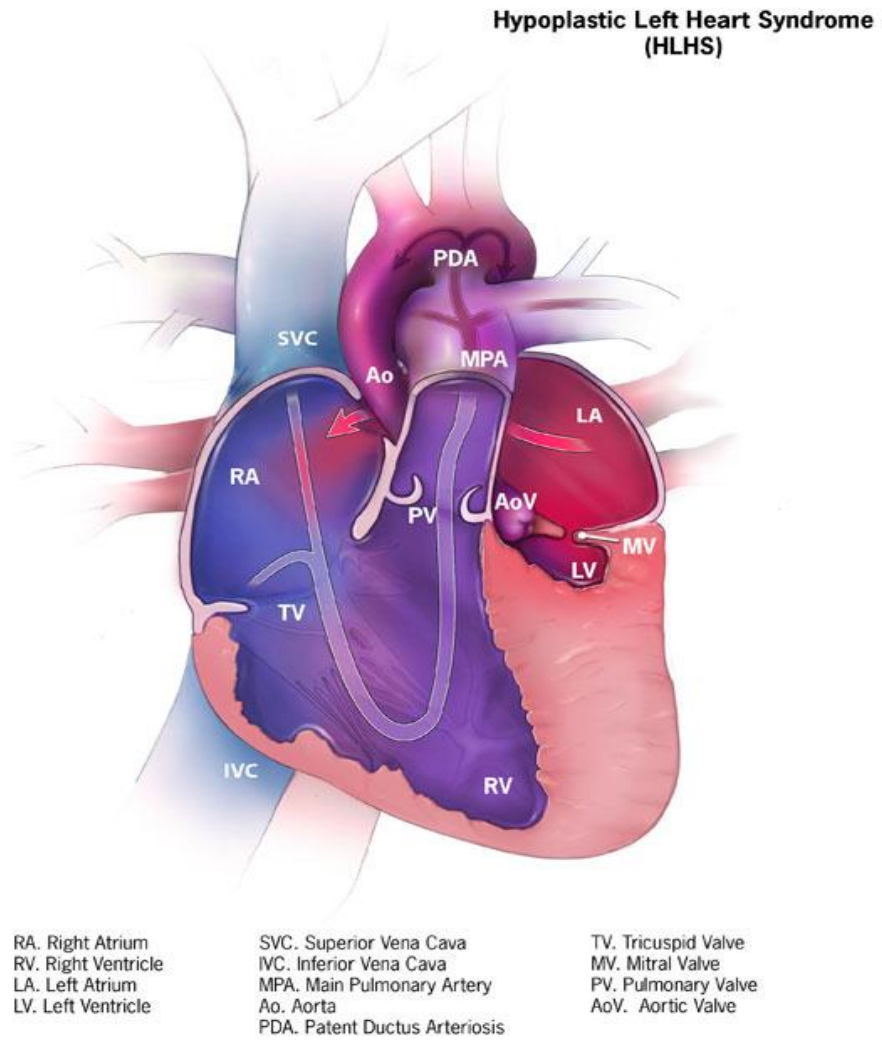
In babies with hypoplastic left heart syndrome, the left side of the heart cannot pump oxygen-rich blood to the body properly. During the first few days of life for a baby with hypoplastic left heart syndrome, the oxygen-rich blood bypasses the poorly functioning left side of the heart through the two openings of the fetal heart (the patent ductus arteriosus and the patent foramen ovale). The right side of the heart then pumps blood to both the lungs and the rest of the body. However, among babies with hypoplastic left heart syndrome, when these openings close, it becomes hard for oxygen-rich blood to get to the rest of the body.

Babies with hypoplastic left heart syndrome might not have trouble for the first few days of life while the patent ductus arteriosus and the patent foramen ovale (the normal openings in the heart) are open, but quickly develop signs after these openings are closed, including:

- Problems breathing,
- Pounding heart,
- Weak pulse, or
- Ashen or bluish skin color.

During a physical examination, a doctor can see these signs or might hear a heart murmur (an abnormal whooshing sound caused by blood not flowing properly). If a murmur is heard or other signs are present, the health care provider might request one or more tests to make a diagnosis, the most common test being an echocardiogram. Echocardiography also is useful for helping the health care provider follow the child's health over time.

Treatments for hypoplastic left heart syndrome include medicines to help strengthen the heart muscle, lower their blood pressure, and help the body get rid of extra fluid; a special high-calorie formula to help babies who tire during feeding to gain weight; and surgery, which do not cure hypoplastic left heart syndrome, but help restore heart function. In some cases, a heart transplant is needed.



This image was provided by the Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities.

- ***Tetralogy of Fallot***

Tetralogy of Fallot (TOF) is a birth defect that affects normal blood flow through the heart. It happens when a baby's heart does not form correctly as the baby grows and develops in the mother's womb during pregnancy.

Tetralogy of Fallot is made up of the following four defects of the heart and its blood vessels (see figure below):

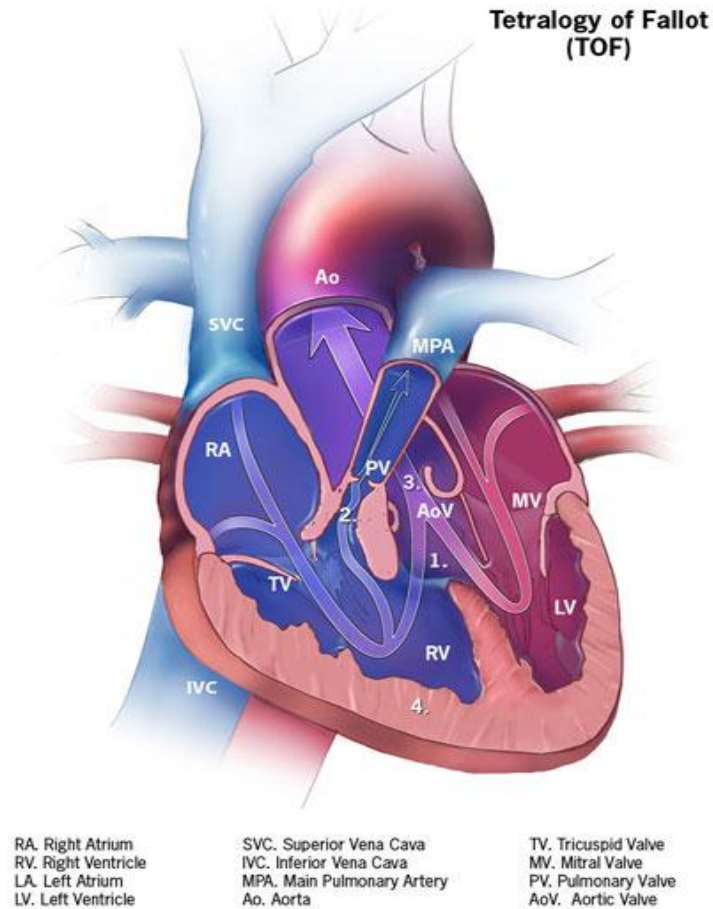
1. A hole in the wall between the two lower chambers (ventricles) of the heart. This condition also is called a ventricular septal defect.
2. A narrowing of the pulmonary valve and main pulmonary artery. This condition also is called pulmonary stenosis.
3. The aortic valve, which opens to the aorta, is enlarged and seems to open from both ventricles, rather than from the left ventricle only, as in a normal heart. In this defect, the aortic valve sits directly on top of the ventricular septal defect.
4. The muscular wall of the lower right chamber of the heart (right ventricle) is thicker than normal. This also is called ventricular hypertrophy.

This heart defect can cause oxygen in the blood that flows to the rest of the body to be reduced. Infants with Tetralogy of Fallot can have a bluish-looking skin color, which is called cyanosis, because their blood doesn't carry enough oxygen. At birth, infants might not have blue-looking skin, but later might develop sudden episodes of bluish skin during crying or feeding. These episodes are called tet spells. Infants with Tetralogy of Fallot or other conditions causing cyanosis can have problems including:

- A higher risk of getting an infection (endocarditis) of the layers of the heart.
- A higher risk of having irregular heart rhythms (arrhythmia).
- Dizziness, fainting, or seizures, because of the low oxygen levels in their blood.
- Delayed growth and development.

Tetralogy of Fallot may be diagnosed during pregnancy or soon after the baby is born. Tetralogy of Fallot usually is diagnosed after a baby is born, often after the infant has an episode of turning blue during crying or feeding (a tet spell). Some findings on a physical exam may make the health care provider think a baby may have Tetralogy of Fallot, including bluish-looking skin or a heart murmur. The health care provider can request one or more tests to confirm the diagnosis. The most common test is an echocardiogram. An echocardiogram is an ultrasound of the heart that can show problems with the structure of the heart and how the heart is working (or not) with this defect. Echocardiography also is useful for helping the doctor follow the child's health over time.

Tetralogy of Fallot can be treated by surgery soon after the baby is born. The surgery will improve blood flow to the lungs and the rest of the body. Most infants will live active, healthy lives after surgery. However, they will need regular follow-up visits with a cardiologist to monitor their progress and check for other health conditions that may develop as they get older.



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- ***Transposition of the Great Arteries***

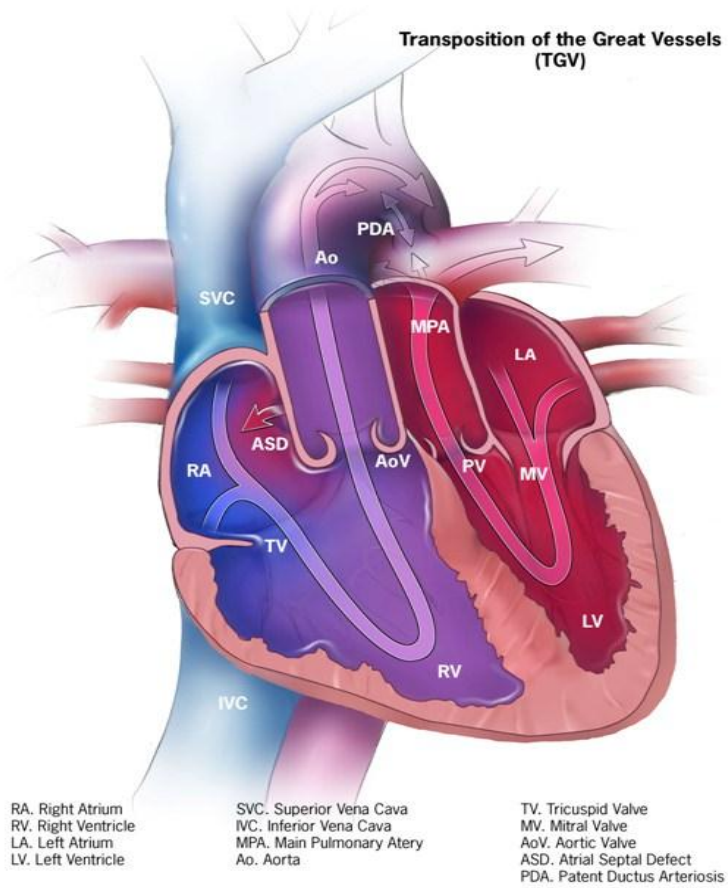
Transposition of the great arteries (TGA) is a heart condition that is present at birth. TGA occurs when the two main arteries going out of the heart—the pulmonary artery and the aorta—are switched in position, or “transposed”.

Normally, blood returning to the heart from the body is pumped from the right side of the heart through the pulmonary artery to the lungs. There, it receives oxygen and returns to the left side of the heart. Then, the oxygen-rich blood is pumped from the left side of the heart through the aorta to the body. In TGA, blood returning from the body bypasses the lungs and is pumped back out to the body. This occurs because the main connections are reversed. The pulmonary artery, which normally carries oxygen-poor blood from the right side of the heart to the lungs, now arises from the left side and carries oxygen-rich blood returning from the lungs back to the lungs. The aorta, which normally carries blood from the left side of the heart to the body, now arises from the right side and carries oxygen-poor blood back out to the body. The result of transposition of these two vessels is that too little oxygen is in the blood that is pumped from the heart to the rest of the body.

Because the main arteries are switched, there are two separate blood circulations instead of a single connected one. Thus, blood with oxygen from the lungs does not get to the rest of the body. This means that TGA is a cyanotic (lacking oxygen) heart defect that leads to a bluish-purple coloring of the skin and shortness of breath.

Symptoms appear at birth or very soon afterwards. How bad the symptoms are depends on whether there is a way for the two separate blood circuits to mix, allowing some oxygen-rich blood to get out to the body.

Surgery might be needed shortly after birth. In most hospitals, a type of surgery called an arterial switch procedure can be used to permanently correct the problem within the first week of life.



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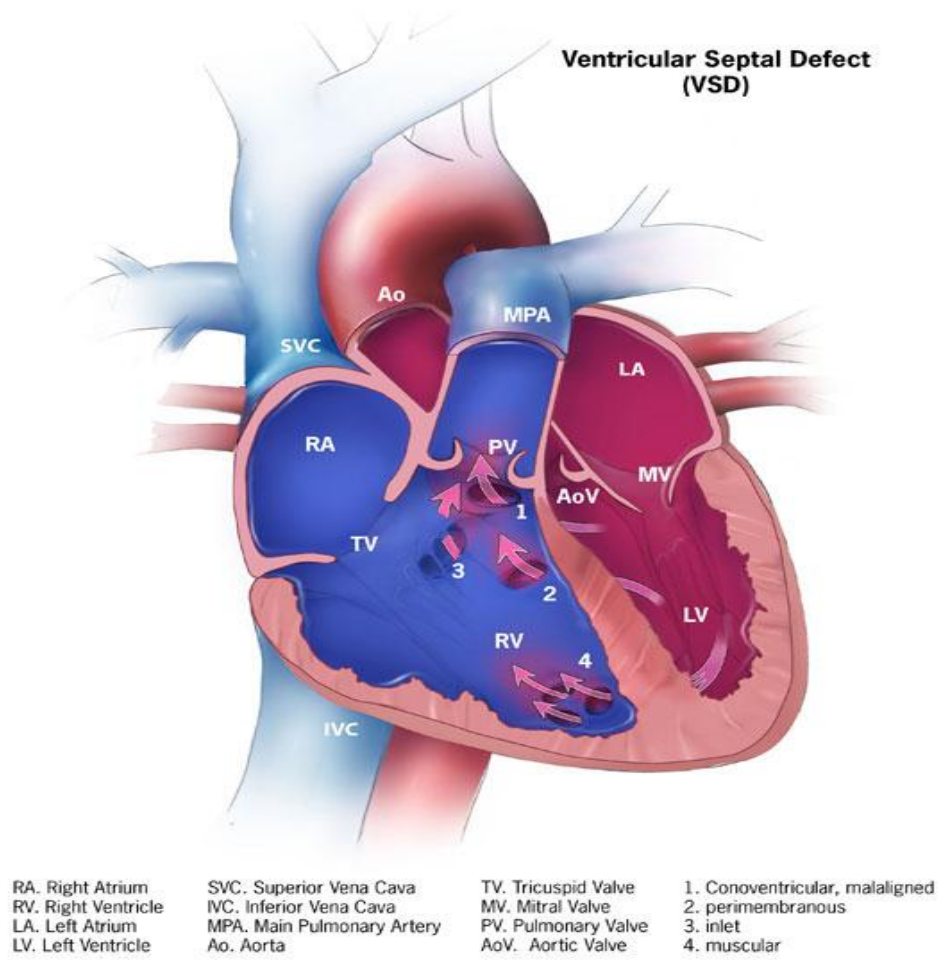
- ***Ventricular Septal Defect***

A ventricular septal defect (VSD) is a birth defect of the heart in which there is a hole in the wall (septum) that separates the two lower chambers (ventricles) of the heart. This wall also is called the ventricular septum. A ventricular septal defect happens during pregnancy if the wall that forms between the two ventricles does not fully develop, leaving a hole. An infant with a ventricular septal defect can have one or more holes in different places of the septum.

In babies with a ventricular septal defect, blood often flows from the left ventricle through the ventricular septal defect to the right ventricle and into the lungs – normally, blood flows from the left ventricle through the aorta to the rest of the body. This extra blood being pumped into the lungs forces the heart and lungs to work harder. Over time, if not repaired, this defect can increase the risk for other complications, including heart failure, high blood pressure in the lungs (pulmonary hypertension), irregular heart rhythms (arrhythmia), or stroke.

A ventricular septal defect usually is diagnosed after a baby is born. The size of the ventricular septal defect will influence what symptoms, if any, are present, and whether a doctor hears a heart murmur during a physical examination. Signs of a ventricular septal defect might be present at birth or might not appear until well after birth. If the hole is small, it usually will close on its own and the baby might not show any signs of the defect. The doctor can request one or more tests to confirm the diagnosis. The most common test is an echocardiogram, which is an ultrasound of the heart that can show problems with the structure of the heart, show how large the hole is, and show how much blood is flowing through the hole.

Treatments for this type of defect depend on the size of the hole and the problems it might cause. Many ventricular septal defects are small and close on their own; if the hole is small and not causing any symptoms, the doctor will check the infant regularly to ensure there are no signs of heart failure and that the hole closes on its own. If the hole does not close on its own, then treatments for ventricular septal defect include medicines to help strengthen the heart muscle, lower their blood pressure, and help the body get rid of extra fluid; a special high-calorie formula to help babies who tire during feeding to gain weight; and surgery. Most children who have a ventricular septal defect that closes (either on its own or with surgery) live healthy lives.



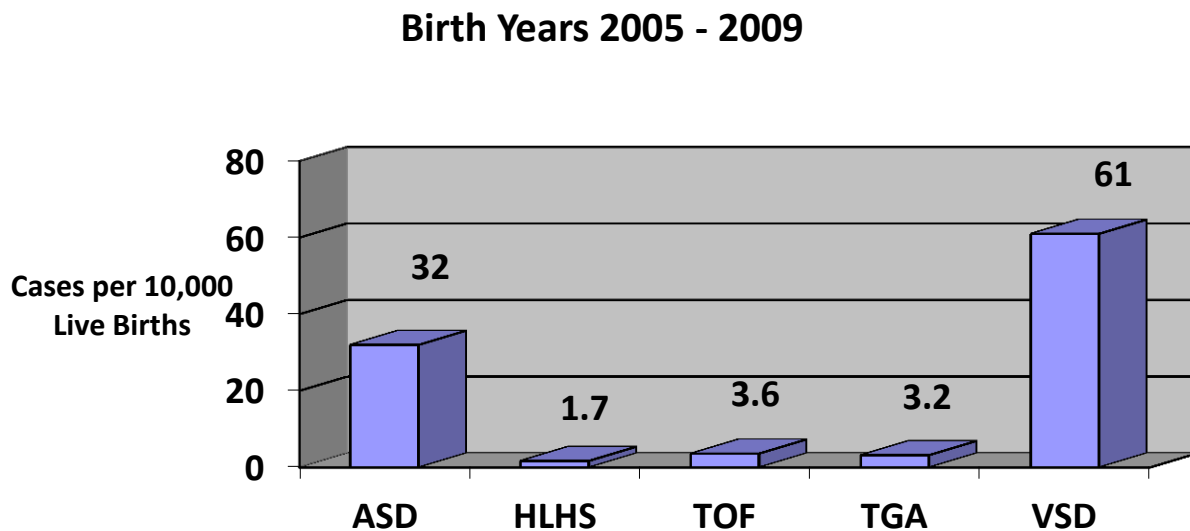
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New Jersey Data:

The Special Child Health Services (SCHS) Registry is a confidential record of infants and children who have birth defects and special health care needs or who are at-risk for developing such needs. Infants and children with a birth defect diagnosed through five years of age are required to be reported to the Registry. A child with a mandated condition identified after 5 years of age through 21 years of age may also be reported but is not mandated by law. While not mandated to be reported, infants and children through 21 years of age with any chronic medical condition, especially those associated with developmental delay, are included in the Registry.

Children may be registered by hospitals, physicians, dentists, audiologists, certified nurse midwives, advanced practice nurses, cytogenetic laboratories and directors of clinical laboratories. SCHS case management units and early intervention service providers may also register. Postmortem examinations resulting in the identification of a mandated condition need to be reported to the Registry.

The figure below shows the number of cases per 10,000 live births of each cardiovascular defect for children born from 2005 through 2009 and registered with the New Jersey SCHS Registry (ASD – atrial septal defect; HLHS – hypoplastic left heart syndrome; TOF – Tetralogy of Fallot; TGA – transposition of the great arteries; VSD – ventricular septal defect).



Resources/Additional Information:

General information related to birth defects:

<http://www.cdc.gov/ncbddd/birthdefects/index.html>

General information congenital heart defects:

<http://www.cdc.gov/ncbddd/heartdefects/index.html>

New Jersey Department of Health information for children with special needs, (Special Child Health Services Registry and services/resources for families):

<http://nj.gov/health/fhs/sch/index.shtml>