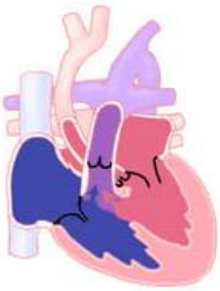
Interrupted Aortic Arch (IAA)

The aorta is the large arch-shaped vessel that brings oxygenated blood out from the top of the heart to the rest of the body. In a child with an **Interrupted Aortic Arch (IAA)**, part of the aortic arch is missing, and blood cannot be delivered normally.

The aorta normally branches out into 3 arteries, sending blood to different parts of the body. In IAA Type B (the most common type of IAA in children with 22q11.2 deletion syndrome), the aorta is interrupted between left carotid and left subclavian artery (the 2nd and the 3rd arterial vessels arising from the aorta). There is a complete obstruction (block) to oxygenated blood outflow from the left ventricle through the aorta, and the lower body does not receive oxygenated blood. Newborn babies typically still have an alternate pathway (**Ductus Arteriosus, DA**) to send blood to the lower body. However, that path closes within hours or days as part of normal development, and the baby becomes very sick from a lack of oxygen. So, in this case, it is **critical that the DA stays patent (unclosed)** to ensure that the oxygenated blood reaches the lower extremities of the body.

Babies who have IAA usually also have a **Ventricular Septal Defect (VSD)**, which result in the mixing of oxygen-rich and oxygen-poor blood. Please see our info sheet on Ventricular Septal Defect for more information.

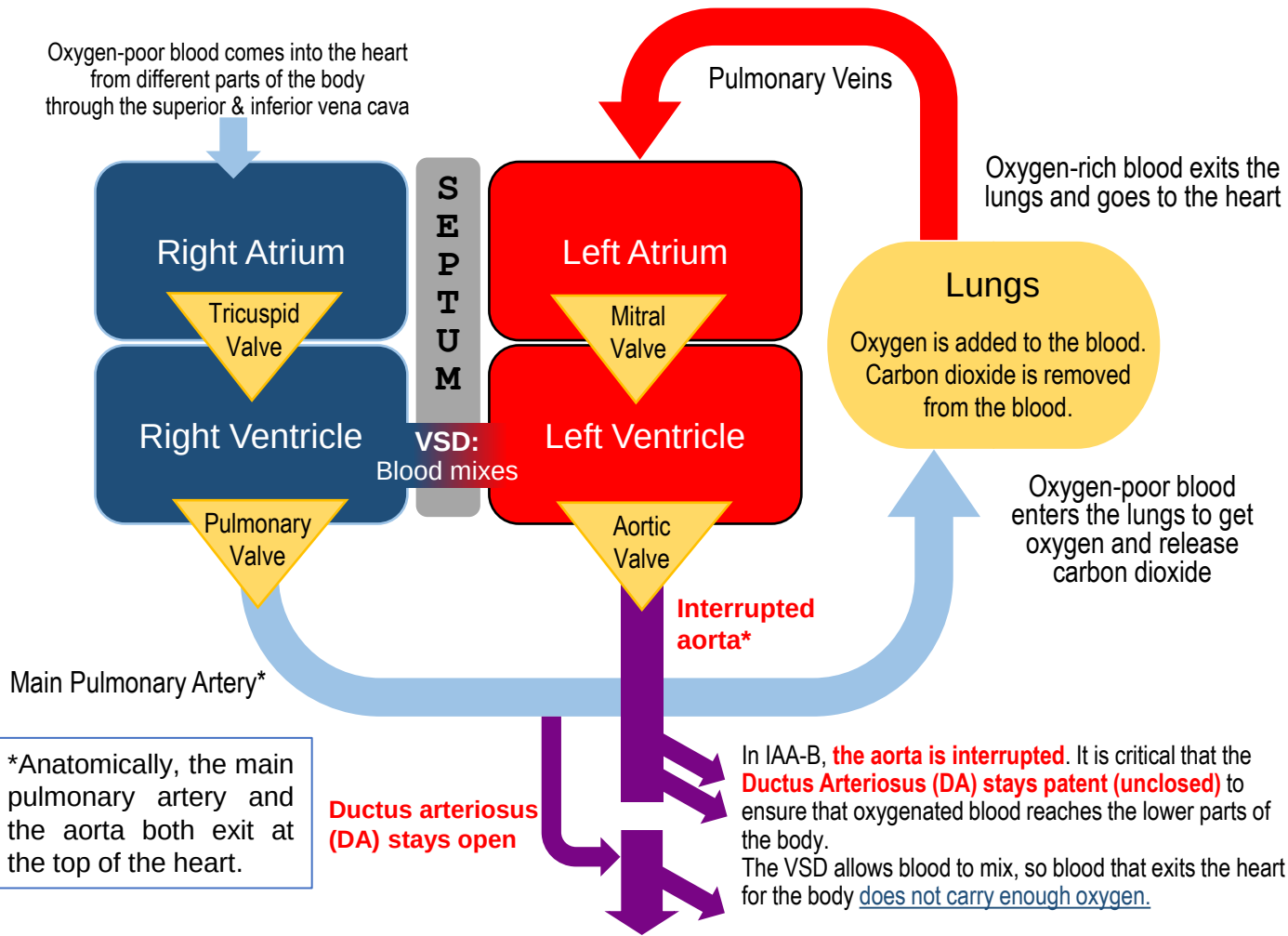


Please click on the image to see a larger version on the website of the Cincinnati Children's Hospital

A Schematic Diagram of the Heart with Interrupted Aortic Arch Type B

Shown as if you are facing the patient; Not to scale

Please also see our schematic diagram of The Heart and Normal Blood Flow for comparison.



Interrupted Aortic Arch (IAA) (continued)

Interrupted Aortic Arch (IAA) and Individuals with 22q Differences

- Of all patients with **Interrupted Aortic Arch Type B (IAA-B)**, 50 to 80% have 22q11.2 deletion syndrome (22q11.2DS).
- 5 to 20% of children with 22q11.2DS are born with IAA.
- In patients with 22q11.2DS, IAA frequently is associated with other complex cardiovascular anomalies.
- In a study, 1 out of 85 children with 22q11.2 duplication syndrome (22q11.2DupS) was born with IAA

IAA and Ductus Arteriosus (DA)

- When a fetus is still in the womb, a connection between the aorta and the pulmonary artery called **Ductus Arteriosus (DA)** lets blood flow from the fetal heart to the lower body. DA exists whether the fetus is normal or have IAA.
- In IAA, when the baby is first born, blood can still travel from the heart to the lower body **only if the DA is patent (staying unclosed)**. Symptoms of IAA may not be obvious right away.
- As the **DA begins to close within the first days of life**, the baby will start to show signs that he/she is not getting enough oxygen. Symptoms include weakness, being very tired, poor feeding, rapid breathing, fast heart rate, and low oxygen levels in the lower parts of the body.

Diagnosis for IAA

- IAA can be diagnosed during pregnancy using fetal **ultrasound** or **fetal echocardiogram**. If IAA is indeed diagnosed before birth, a plan can be made for delivery and care.
- After the baby is born, IAA can be diagnosed using **echocardiogram**. Other examinations such as MRI or CT may be useful for diagnostic confirmation and preoperative management.
- Once symptoms appear, it is critical to stabilize the baby right away.

Treatment for IAA

- The medical provider may use medications that are essential **to keep the DA (the alternate pathway) open**, so that blood can flow from the baby's heart to his/her lower body until surgery can be done.
- Additional treatments and medications may be given to help the baby breathe better, get rid of excess fluids, get the heart to pump better, adjust levels of blood gasses, and get nutrition, etc.
- **Surgery** must be done to connect the aorta back together and to patch up the VSD.

Ongoing care for IAA

1. Regular **follow-up with a cardiologist (with expertise in congenital heart defects)** is necessary. The cardiologist may recommend **tests** to make sure the heart works well.
2. Additional medications and surgeries may be needed.
3. Some patients may need to **limit their activities**. Please discuss exercise tolerance with your cardiologist.
4. Some people may need **endocarditis prophylaxis** – This means taking antibiotics before certain dental procedures to prevent bacteria from causing infections in the heart. Please discuss with your cardiologist.
5. Women planning on **getting pregnant should discuss their risks** with their healthcare providers and cardiologist.

For more info, please visit the websites of the Cincinnati Children's Hospital and the Children's Hospital of Philadelphia.

22q International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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