Aortic Valve Abnormalities

From the Simple to the Complex

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Aortic Valve Abnormalities

- Aortic Valve Abnormalities are the most common valve abnormalities seen in the general population.

- It is estimated that 1% of the general population has such an abnormality.

- 50% of adult patients with aortic valve disease are thought to be congenital in origin.

- In children LVOT obstructive lesions account for 6% of all CHD in children with an incidence of about 4/10,000 births. 70-80% of these are male.
Why Is the Aortic Valve Abnormal?

- Theories relate to morphogenesis, or flow dynamics.
- The aortic valve is formed by the 31-35 day of gestation.
- Most studies suggest that the abnormality is genetic in origin.
Not Statistically Relevant Fact

• A completely biased non-randomized sampling of the children of the Children’s Heart Center physicians showed that 8% of them have an aortic valve abnormality.
Aortic Valve Abnormalities

- A normal aortic valve has 3 leaflets, and when closed from above looks like the Mercedes Benz Emblem.

- Most aortic valve abnormalities result from fusion of two of the cusps together, making the valve a bi-leaflet (bicommissural) valve.

- Very few of the aortic valve abnormalities are unicuspid valves.
Bi-leaflet Aortic Valves

- The majority of abnormal aortic valves, 70%, have fusion between the right and left coronary cusps with the leaflets oriented anterior-posterior.

- The second most common abnormality is fusion between the right and non-coronary cusps with the leaflets oriented right and left.

- The least common variant is fusion of the left and non-coronary cusp.
Normal tricuspid aortic valve

Position of aortic valve in the heart

Bicuspid aortic valve

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Beyond the 2-Leaflet Aortic Valve

• There are unicuspid aortic valves where 2 out of 3 commissures are fused, with the opening of the valve leftward and posterior to the valve annulus.

• Absence of all 3 commissaries results in aortic atresia (HLHS).
Which Leaflets Are Fused Has Clinical Relevance

- Patients with fusion of the right and non coronary commissure have worse aortic stenosis and insufficiency. They also have a more rapid progression of their disease.

- Patients with the more common type, where the right and left commissures are fused, have less degree of valvular disease but have a higher risk of coarctation of the aorta.
Complications from Aortic Valve Abnormalities

- Some patients have no signs or symptoms from their valve abnormality.

- Some patients will have a dilated ascending aorta—thought to be due to flow dynamics or an intrinsic abnormality in the aorta itself.

- Patients can have varying degrees of stenosis and/or insufficiency of their valves.
Other Associations with Aortic Valve Abnormalities

- Mitral valve stenosis is usually seen with general hypoplasia of left heart structures.

- Coarctation of the aorta accounts for 4-6% of all congenital heart disease with a reported prevalence of 4/10,000 live births. Approximately 80% of patients with coarctation have an aortic valve abnormality.

- Hypoplastic Left Heart Syndrome which is 2-3% of all congenital heart disease, with approximately 2-3 cases/10,000 live births.
PDA (Patent Ductus Arteriosus)
Hypoplastic aorta
Hypoplastic mitral and aortic valves
Hypoplastic left ventricle

PFO (Patent Foramen Ovale)
BAV to HLHS

- Some studies point to flow dynamics secondary to defective valvulogenesis or other elements of cardiovascular system development.
- Theoretically decreased flow across the aortic valve in-utero may result in underdevelopment of the structures on the left side of the heart while increasing flow across the ductus arteriosus.
- For Coarctation there are theories about overly invasive ductal tissue.
A NOT Statistically Relevant Fact

- A Completely Biased Non-Randomized Survey of Children’s Heart Center Physicians showed that 0.8% of our children have an aortic valve abnormality.
Genetic Data

- Most compelling data is related to familial occurrence of aortic valve abnormalities.

- Studies quote a 4-10% recurrence rate of aortic valve abnormalities in 1st degree relatives.

- In 2009 the AHA/ACC recommended screening all first degree relatives of patients with aortic valve abnormalities for that reason.
Genetics

- Family data suggests the genetic link because of the high percent of first degree relatives with aortic valve disease.

- Some studies suggest autosomal dominant inheritance but other studies suggest there are mutations of diverse genes with dissimilar inheritance patterns.

- Several syndromes associated with aortic stenosis and coarctation including Turner syndrome and Jacobsen’s syndrome.
Conclusions

- Aortic Valve Abnormalities are very common and often not diagnosed until adulthood.

- Family studies suggest that this is a genetically transmitted disease.

- Although a BAV is the most common, there are many families with a 1st degree relative with any LVOTO, including HLHS and coarctation.
Conclusions

- Screening of 1st degree relatives recommended because of high recurrence of up to 10% in first degree relatives.

- Screening can help predict and maybe prevent complications from aortic valve disease.