

Aortic stenosis and atresia

Congenital obstruction of the left ventricular outflow tract

Recognize where obstruction occurs, how cardiac structure responds,
and why ductal patency matters in severe neonatal disease.

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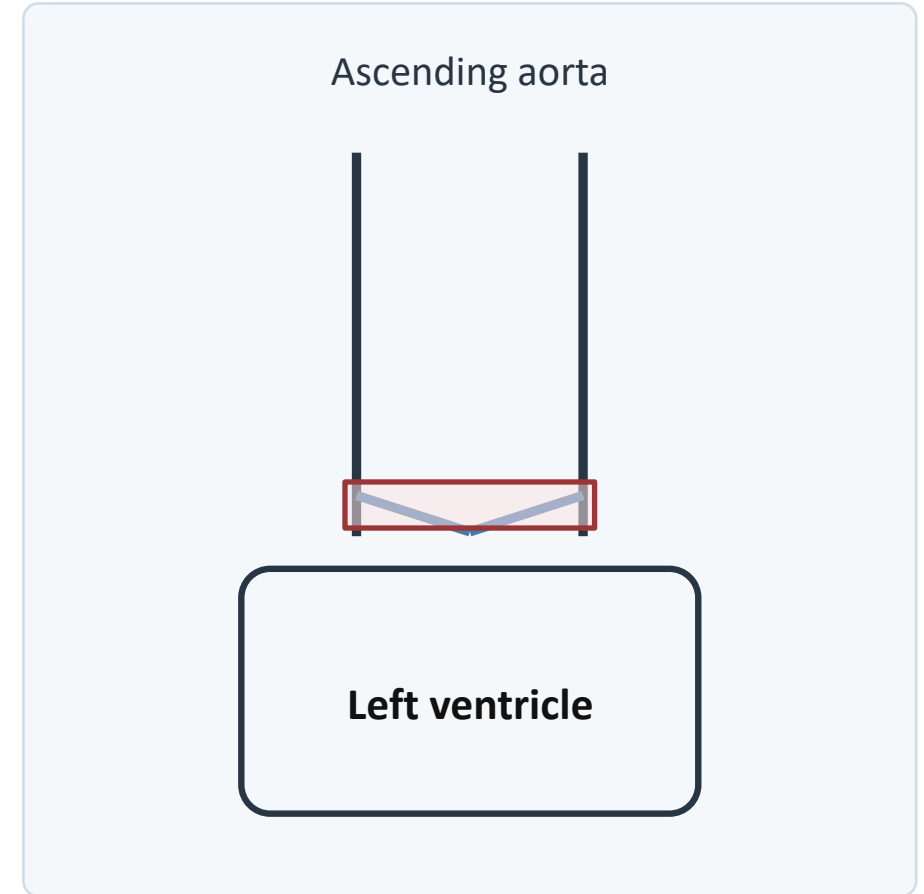
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Learning objectives

- Localize congenital obstruction to the valve, below the cusps, or above the valve.
- Relate valvular morphology to impaired systolic ejection and ventricular pressure load.
- Describe the severe stenosis–atresia–hypoplastic left heart spectrum.
- Explain the importance of the patent ductus arteriosus and compare the three anatomic levels.

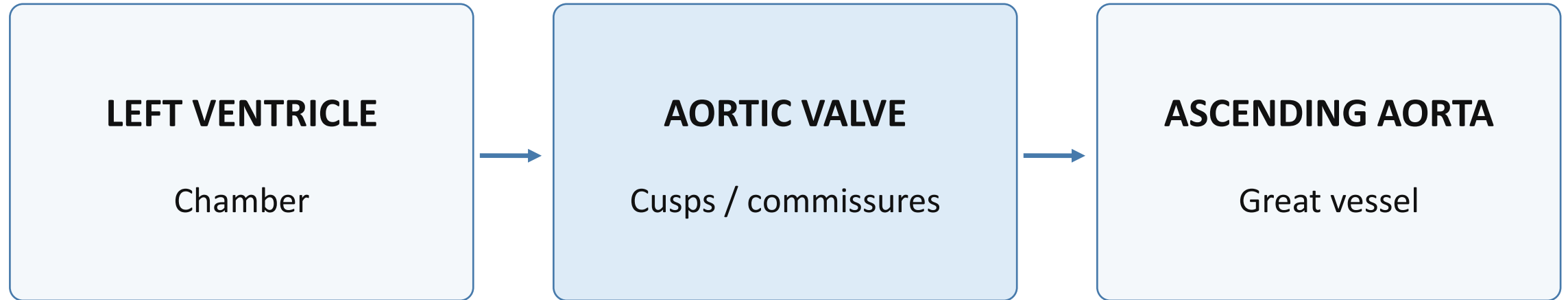
Aortic outflow disease belongs to obstructive CHD

- Congenital heart disease comprises structural abnormalities present at birth.
- Obstructive lesions narrow a chamber, valve, or great vessel; Complete developmental obstruction is atresia.
- Aortic stenosis affects the left ventricular outflow route into the systemic circulation.
- Shunts and obstructions are distinct hemodynamic categories, although complex defects may coexist.



Anatomic route: Ventricle → valve → aorta

The site of a fixed narrowing determines which structure is malformed.



Schematic: Direction of normal antegrade ejection

Three distinct levels of narrowing

All three impair the left ventricular outflow pathway, but the primary tissue lesion differs.

SUPRAVALVULAR

Ascending aorta

VALVULAR

Valve cusps

SUBVALVULAR

Subaortic collar

Stenosis and atresia are not synonyms

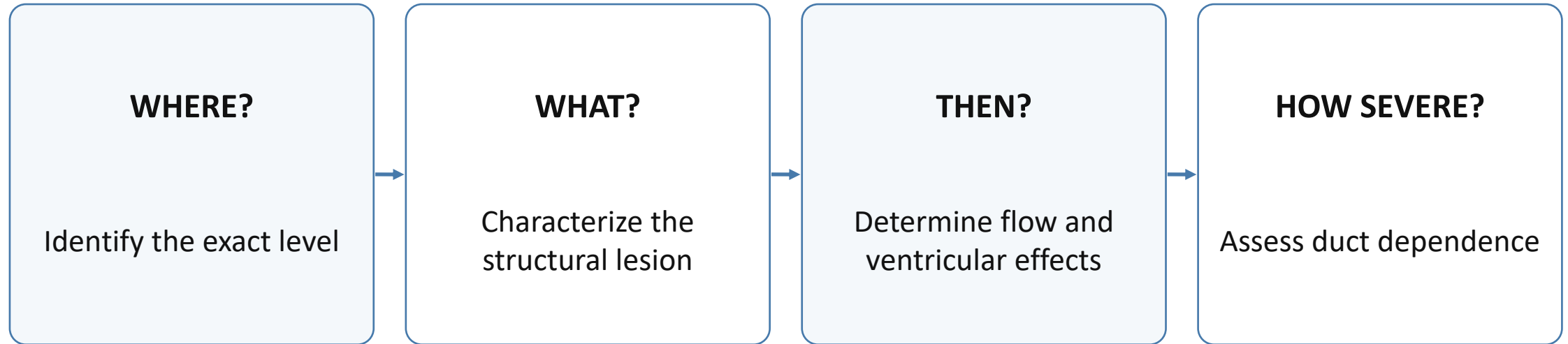
Stenosis

- An outflow lumen remains, but is narrowed.
- Blood can traverse the affected segment.
- Consequences depend strongly on severity.

Atresia

- The outflow orifice is completely obstructed.
- Antegrade flow across that level is absent.
- Systemic output may require an alternative route.

The fundamental pathology question



Causal sequence / anatomic reasoning schematic

Most congenital aortic valve stenosis is isolated

- The valvular lesion as isolated in 80% of cases.
- “Isolated” describes the lesion’s association status, not the severity of obstruction.
- Severe left ventricular outflow obstruction can instead form part of hypoplastic left heart syndrome.
- Anatomic context is essential before predicting clinical behavior.

Anatomic reasoning

Isolated valve lesion

The presence or absence of associated malformations must be assessed.

Schematic

Aortic stenosis among congenital malformations

Selected lesion	Incidence per million live births	Share of tabulated defects
Aortic stenosis	388	4%
Pulmonary stenosis	836	8%
Coarctation of the aorta	492	5%

Values are textbook table entries; The underlying table excludes bicuspid aortic valves.

Congenital defects reflect cardiac morphogenesis

- Major cardiovascular structures form during embryonic weeks 3–8.
- Cardiac progenitors contribute to chambers, outflow tracts, and supporting tissues.
- Developmental errors can be localized to one valve or involve a broad segment of the left heart.
- Anatomic severity at birth reflects developmental consequences, not merely postnatal remodeling.

Anatomic reasoning

Embryonic morphogenesis

A fetal structural lesion can affect later chamber growth.

Schematic

Etiology: Avoid assuming one causal gene

- CHD may reflect genetic and environmental influences on cardiac morphogenesis.
- The broad CHD chapter emphasizes that the precise cause is unknown in most cases.
- A named molecular association is not established for every valvular stenosis.
- The defined genetic association in this topic concerns some supra-ventricular lesions and elastin.

Anatomic reasoning

Molecular interpretation

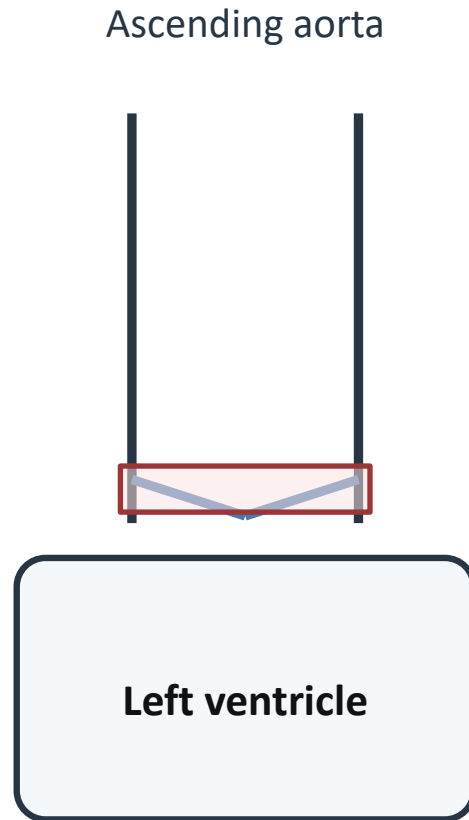


Only report causal associations supported by the source.

Schematic

Valvular obstruction is a cusp-level lesion

The obstruction resides at the moving cusps, not in an independent wall segment.

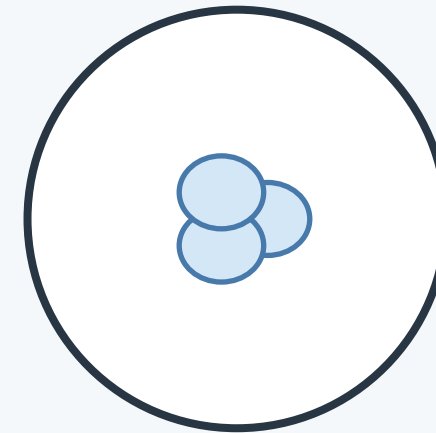


- Valvular morphology varies among affected hearts.
- Altered cusps or commissures can restrict the opening.
- This is distinct from a fibrous ring below the cusps.

Hypoplastic cusps: Insufficient cusp development

- Hypoplastic aortic cusps are abnormally small.
- Cusp size is an anatomic descriptor, not a measured severity grade in the source.
- The essential diagnostic focus is restricted valvular anatomy and impaired outflow.
- A schematic cannot replace gross specimen inspection.

Aortic cusp morphology



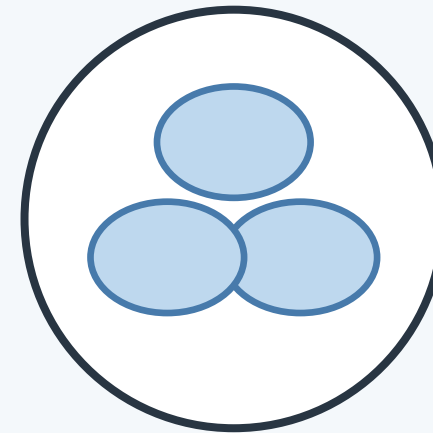
Small cusps

Schematic · not a specimen

Dysplastic cusps: Thickened and nodular

- Dysplasia produces thickened, nodular aortic valve cusps.
- This is a morphologic description of congenital tissue abnormality.
- Cusp thickening can contribute to reduced effective orifice opening.
- Do not infer a particular microscopic composition without source evidence.

Aortic cusp morphology



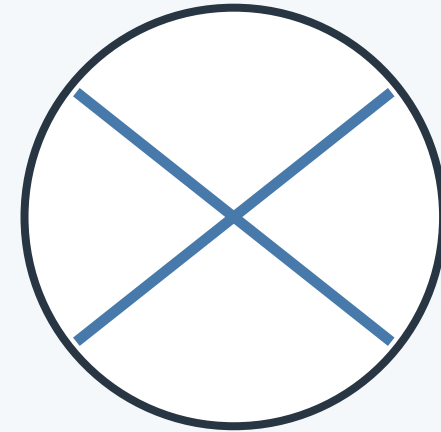
Thickened, nodular cusps

Schematic · not a specimen

Commissural abnormalities alter valve opening

- Congenital valves can be abnormal in the number of commissures.
- Valves usually having one commissure or none among these anomalies.
- Commissures define cusp separation and functional opening geometry.
- Commissural abnormalities are distinct from cusp hypoplasia and dysplasia, though they may coexist.

Aortic cusp morphology



Altered commissural pattern

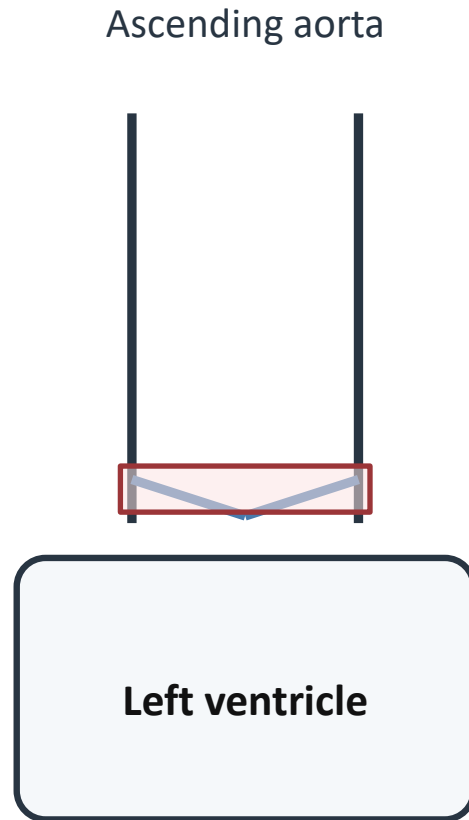
Schematic · not a specimen

Valvular morphology can be heterogeneous

Description	Primary abnormality	What it changes
Hypoplastic	Small cusps	Cusp development
Dysplastic	Thickened, nodular cusps	Cusp architecture
Abnormal commissures	One or no commissures	Opening configuration

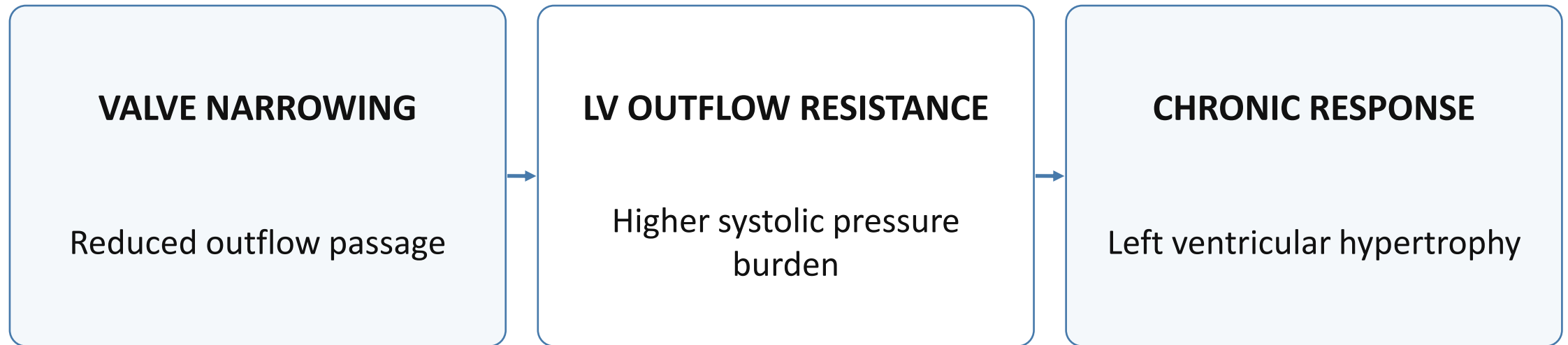
What becomes obstructed during systole?

A small effective outlet restricts blood ejection from the left ventricle into the aorta.



- Ventricular ejection requires passage through the valve.
- Narrowing increases resistance to forward flow.
- The obstruction is anatomically upstream of the systemic arteries.

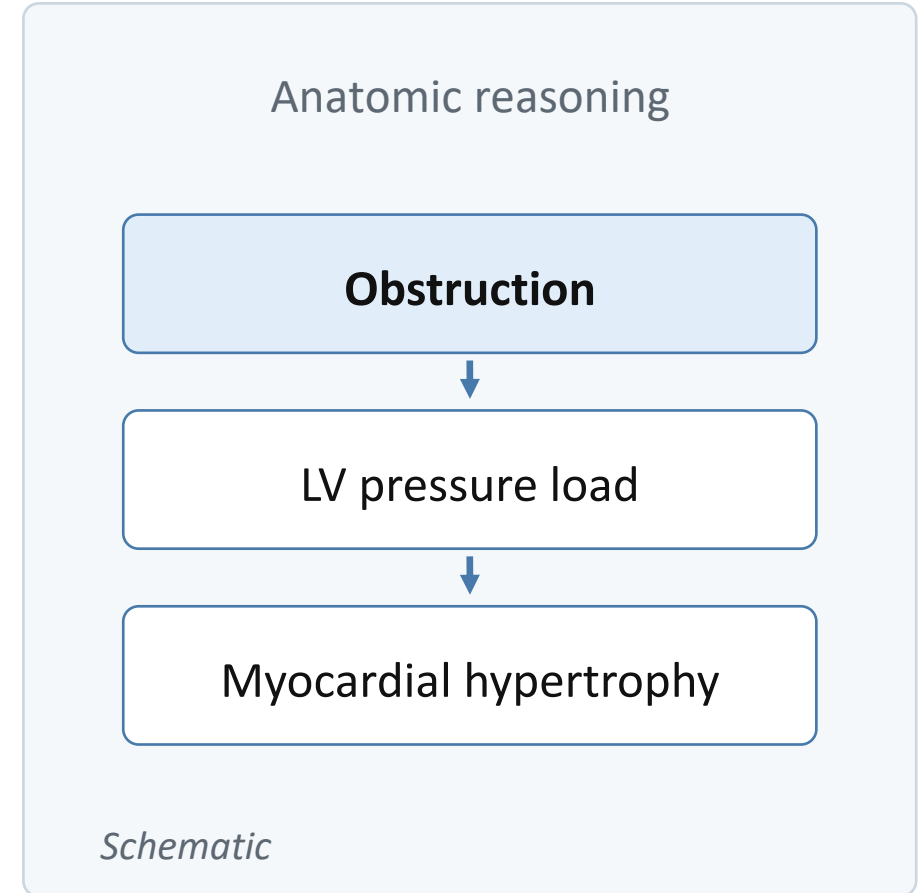
A pressure load develops proximal to obstruction



Causal sequence / anatomic reasoning schematic

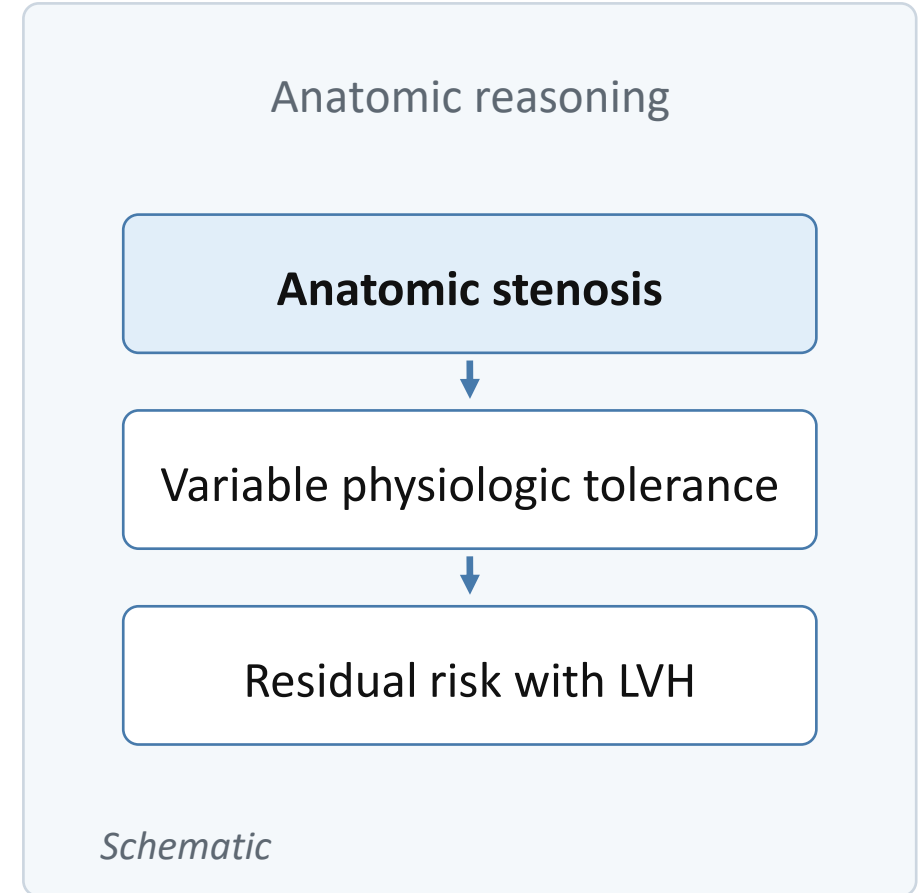
Pressure hypertrophy is a ventricular response

- Robbins explicitly associates congenital obstruction with left ventricular pressure hypertrophy.
- The myocardium adapts to a sustained pressure burden by increasing muscle mass.
- Hypertrophy is a response to loading, not a correction of the malformed valve.
- Do not confuse acquired hypertrophy with prenatal chamber hypoplasia.



Obstruction may be tolerated despite hypertrophy

- Unless stenosis is very severe, it is generally well tolerated.
- Clinical tolerance does not mean the left ventricle remains structurally normal.
- Pressure hypertrophy may still develop despite limited symptoms.
- Severe ventricular remodeling carries a risk of sudden cardiac death.



Why a systolic murmur can occur

- During systolic ejection, blood moves across a narrowed outflow pathway.
- The resulting disturbed flow can produce an audible systolic murmur.
- Robbins specifically highlights a prominent systolic murmur in subaortic stenosis.
- A murmur suggests turbulent flow; It does not identify the exact level by itself.

Anatomic reasoning

Systolic ejection



Disturbed flow

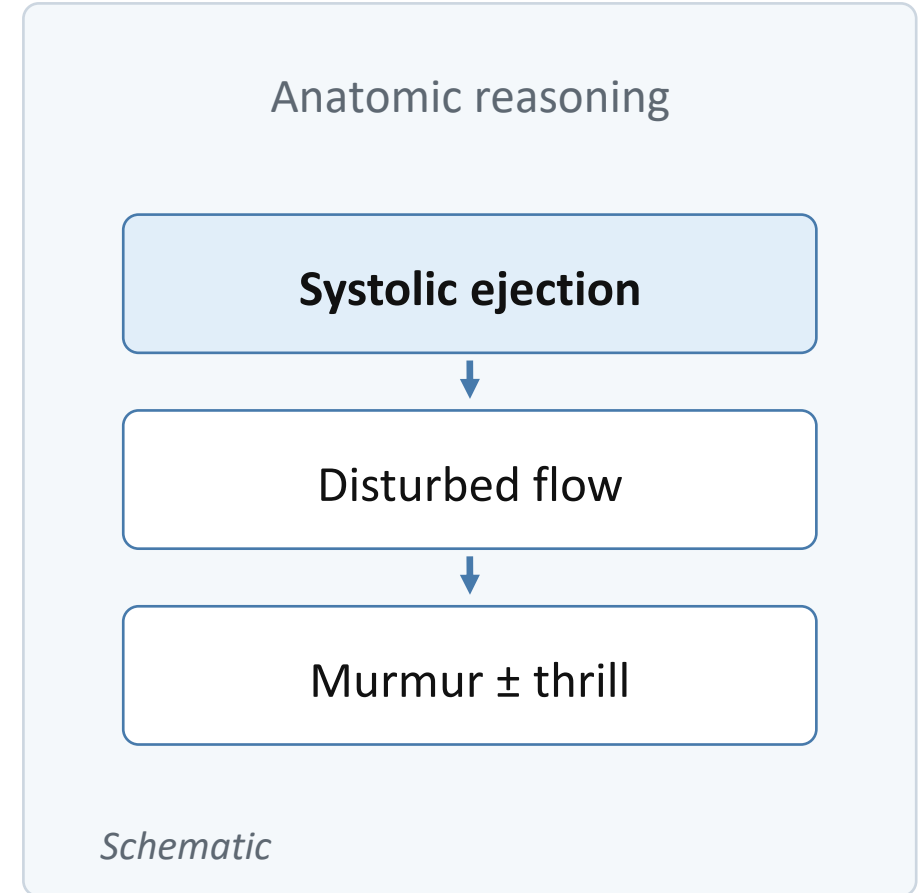


Murmur ± thrill

Schematic

A thrill implies strong palpable flow disturbance

- A thrill is a palpable vibration accompanying a forceful murmur.
- Subaortic stenosis can have a thrill according to the textbook.
- The examination finding reflects flow disturbance rather than histologic subtype.
- Define the anatomic obstruction separately from its clinical sign.



Anatomic severity changes the consequence

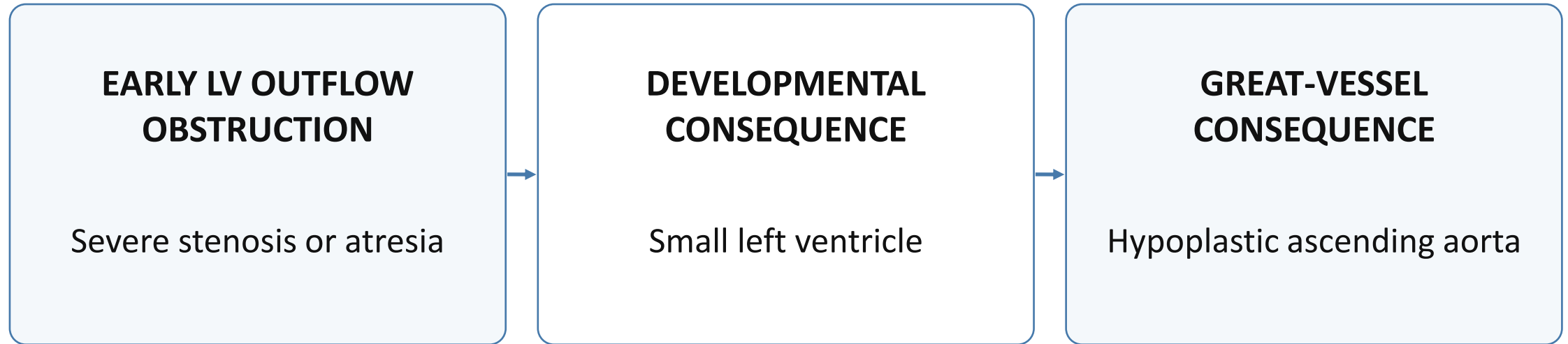
Less severe stenosis

- A residual outflow channel is present.
- Symptoms may be absent or limited.
- Chronic left ventricular pressure load is still possible.

Critical obstruction / atresia

- Forward aortic outflow is profoundly restricted or absent.
- Developmental left-heart hypoplasia may coexist.
- Maintenance of systemic flow can become duct-dependent.

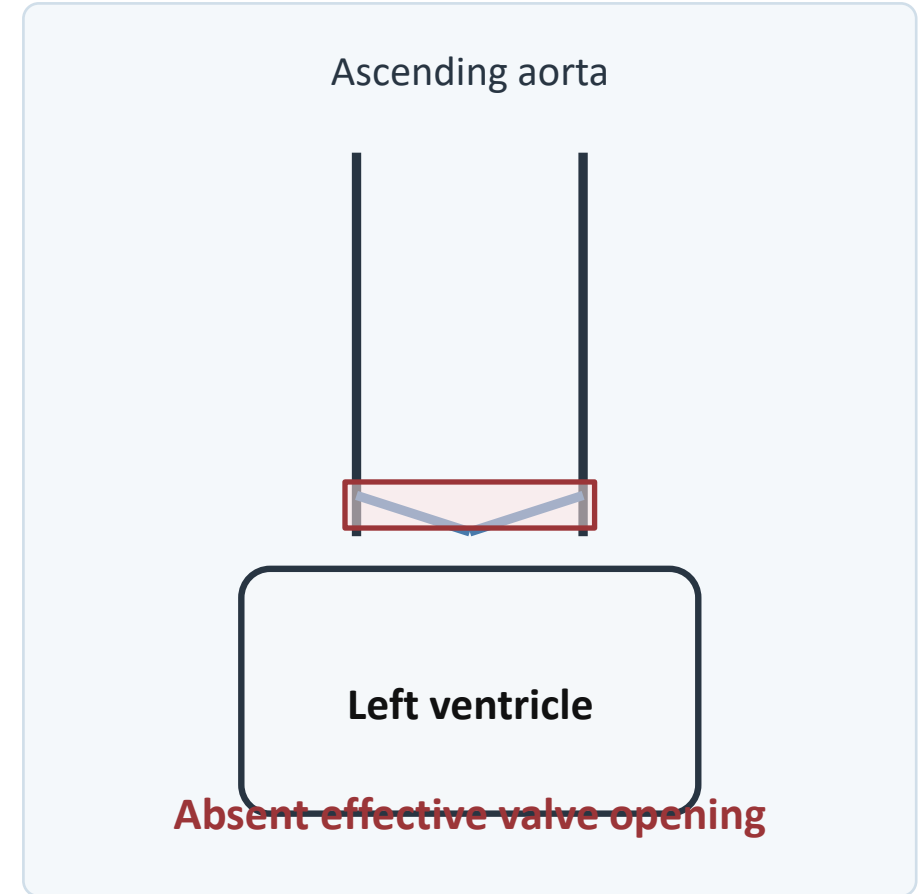
Severe congenital obstruction can change growth



Causal sequence / anatomic reasoning schematic

Atresia abolishes normal valve-level ejection

- Aortic valvular atresia is complete obstruction at the aortic valve level.
- Blood cannot pass antegrade from the left ventricle across an atretic valve.
- The circulation must rely on an alternative route to supply the aorta.
- In the severe constellation, the patent ductus arteriosus supplies that route.



Prenatal hypoplasia differs from later atrophy

Hypoplasia

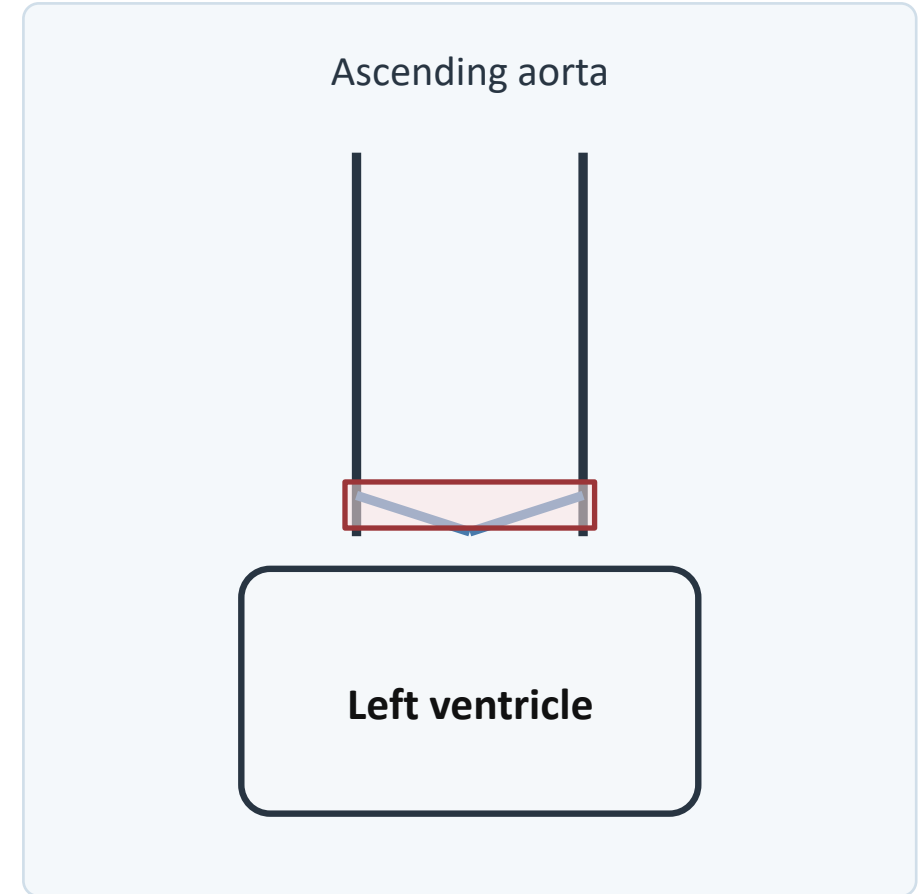
- Reduced chamber volume and muscle mass develop before birth.
- Represents insufficient development or growth.
- Severe left ventricular outflow disease may produce it.

Atrophy

- A decrease in chamber mass developing after birth.
- A different temporal/pathologic category.
- Should not be substituted for congenital LV hypoplasia.

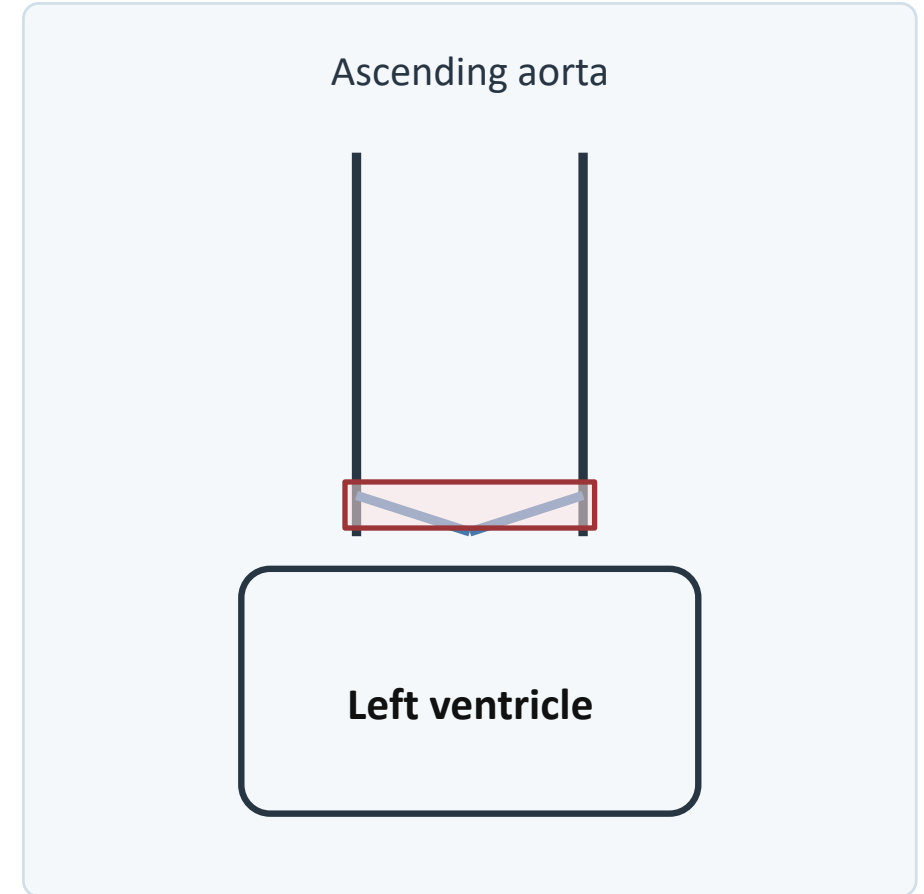
The hypoplastic left ventricle

- Severe congenital aortic stenosis or atresia may produce a small left ventricle.
- An inadequate left ventricular chamber cannot be assumed to provide normal systemic output.
- The abnormality involves the chamber, not just the orifice.
- This is a major step toward the hypoplastic left heart syndrome phenotype.



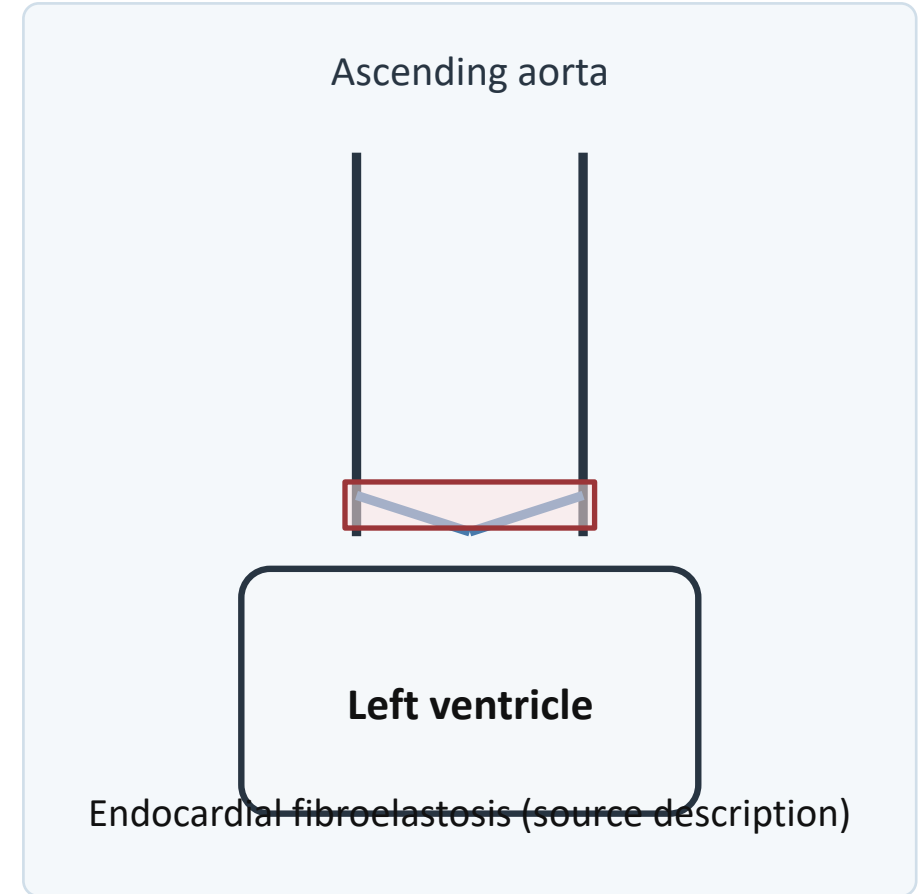
The ascending aorta may also be hypoplastic

- The ascending aorta can be underdeveloped along with the left ventricle.
- Aortic caliber matters because the vessel must deliver flow to the upper systemic circulation and coronary origins.
- Valve obstruction and great-vessel hypoplasia form a linked severe morphology.
- The exact extent of the hypoplasia is not specified in the excerpt.



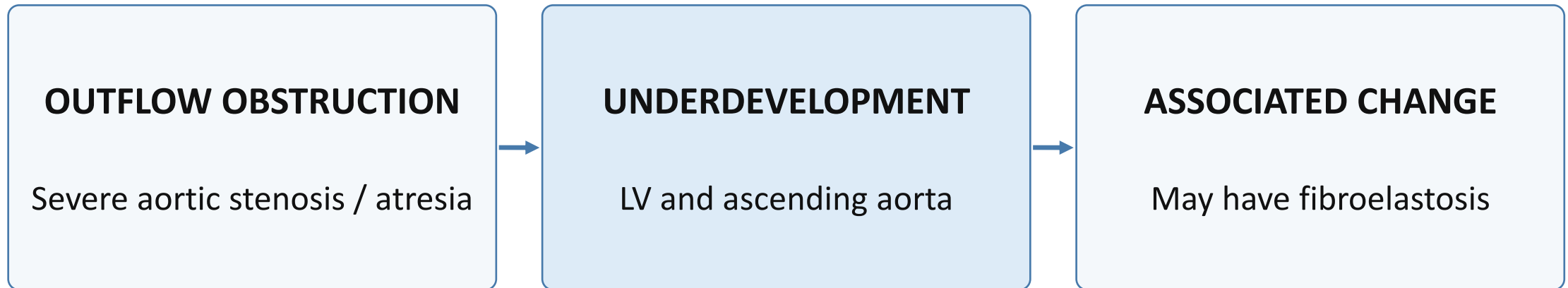
Endocardial fibroelastosis is an associated lesion

- Severe cases may exhibit dense left ventricular endocardial fibroelastosis.
- A striking porcelain-like endocardial appearance.
- This is an associated morphologic finding, not required in every case.
- Do not present a synthetic photomicrograph as evidence of this lesion.



Hypoplastic left heart: An anatomic constellation

The syndrome designation reflects a combination of severe structural abnormalities.



- Severe aortic stenosis or atresia
- Hypoplastic left ventricle
- Hypoplastic ascending aorta; Fibroelastosis may coexist

Conceptual schematic; Actual anatomy varies

Not every congenital aortic stenosis is HLHS

Isolated valvular stenosis

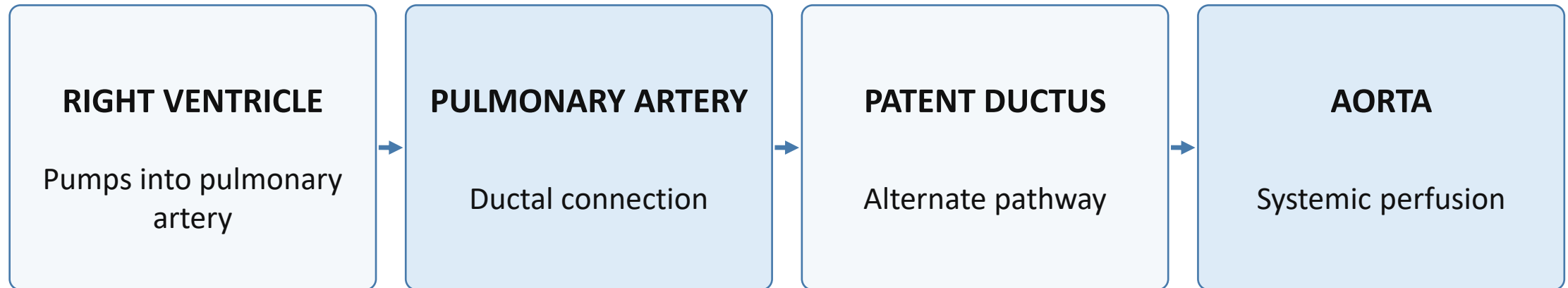
- A focal congenital valve lesion.
- Can be well tolerated when not very severe.
- Does not automatically imply chamber hypoplasia.

Hypoplastic left heart syndrome

- Severe obstructive left-heart malformation.
- Includes underdeveloped LV and ascending aorta.
- Systemic/coronary flow depends on ductal patency description.

The ductus becomes a systemic lifeline

In severe aortic atresia/HLHS, the patent ductus arteriosus permits blood to reach the aorta.

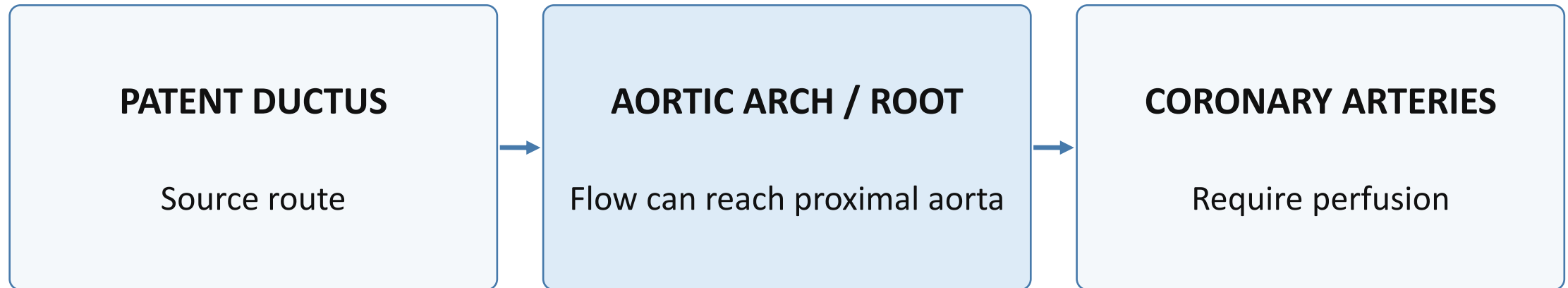


- The ductus connects pulmonary arterial blood flow with the aorta.
- This bypasses the unusable native left ventricular outflow route.
- The systemic circulation becomes vulnerable to ductal closure.

Conceptual schematic; Actual anatomy varies

Why the coronary arteries also depend on flow

Ductal patency is needed to supply both the aorta and coronary arteries.



- The coronary ostia arise from the proximal aortic root.
- In the severe atretic pathway, systemic supply can reach the arch/root by the duct-dependent route.
- Loss of the duct removes a critical route for coronary as well as systemic perfusion.

Conceptual schematic; Actual anatomy varies

Ductal patency and ductal closure

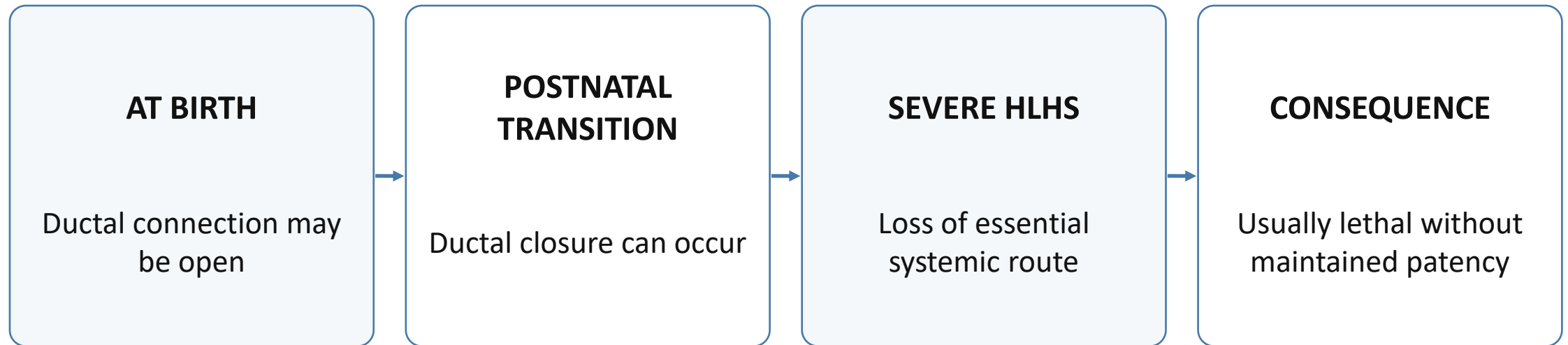
Patent ductus

- An alternative pathway remains open.
- Aorta and coronary circulation can receive blood.
- Severe obstruction may still be physiologically precarious.

Ductus closes

- The alternative pathway is lost.
- Systemic and coronary perfusion may collapse.
- Closure in week 1 as generally lethal in this syndrome.

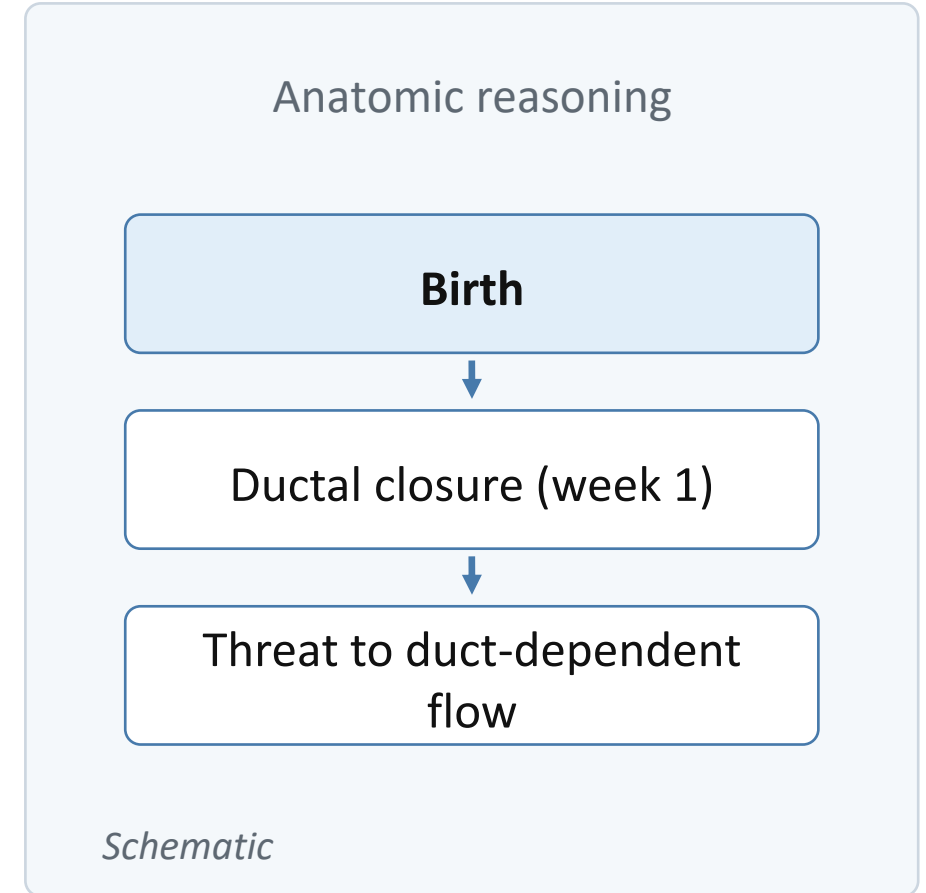
The critical transition occurs after birth



Causal sequence / anatomic reasoning schematic

The first week is a decisive clinical window

- Duct closure during the first week of life as generally lethal in HLHS.
- The mechanism is loss of blood supply to the aorta and coronary arteries.
- This mortality statement applies to the duct-dependent severe constellation—not every congenital aortic stenosis.
- Avoid equating “isolated valvular stenosis” with neonatal duct dependence.



Clinical interpretation starts with duct dependence

- First, decide whether there is a residual route for left ventricular ejection.
- Second, recognize whether the left ventricle and ascending aorta are adequately developed.
- Third, ask whether the patent ductus supplies essential systemic blood flow.
- A change in ductal patency can change physiology even when valve anatomy is unchanged.

Anatomic reasoning

Outflow patency?



Left-heart hypoplasia?

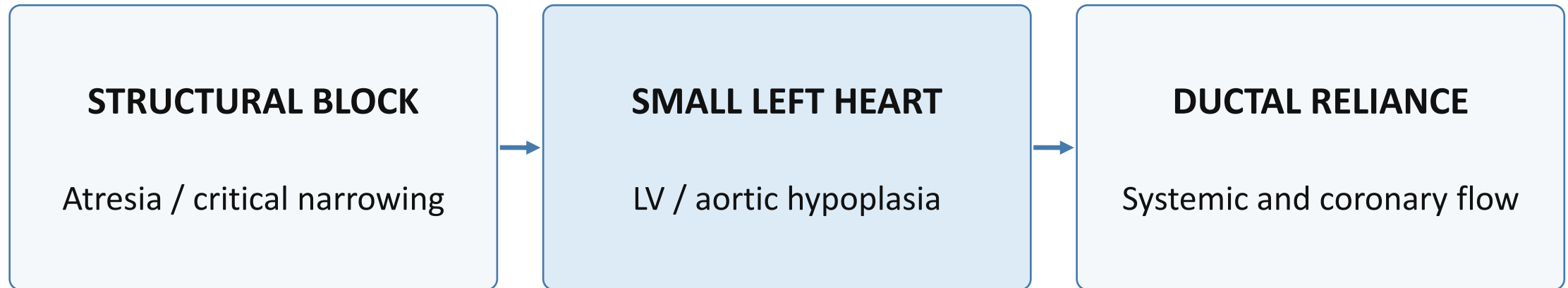


Duct needed?

Schematic

Integrating severe aortic obstruction

The risk arises from combined obstruction, underdevelopment and loss of the alternative circulation.

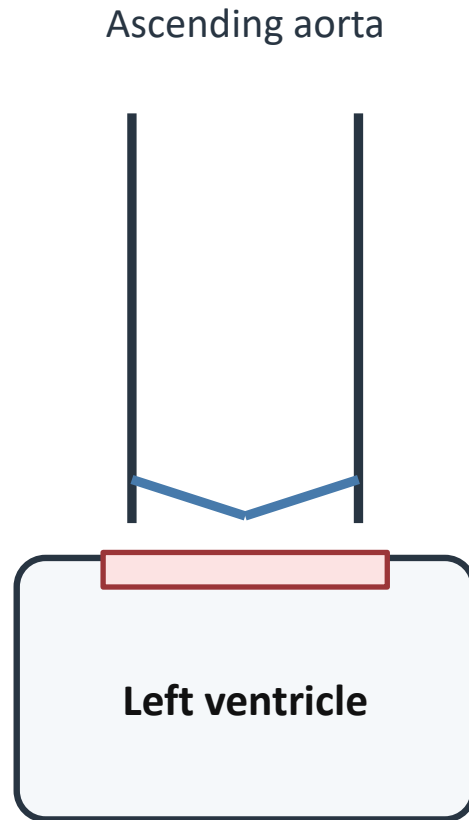


- Aortic obstruction → reduced antegrade route
- LV / ascending aorta hypoplasia → limited native systemic pathway
- PDA-dependent perfusion → hazard of ductal closure

Conceptual schematic; Actual anatomy varies

Subaortic stenosis lies below the cusps

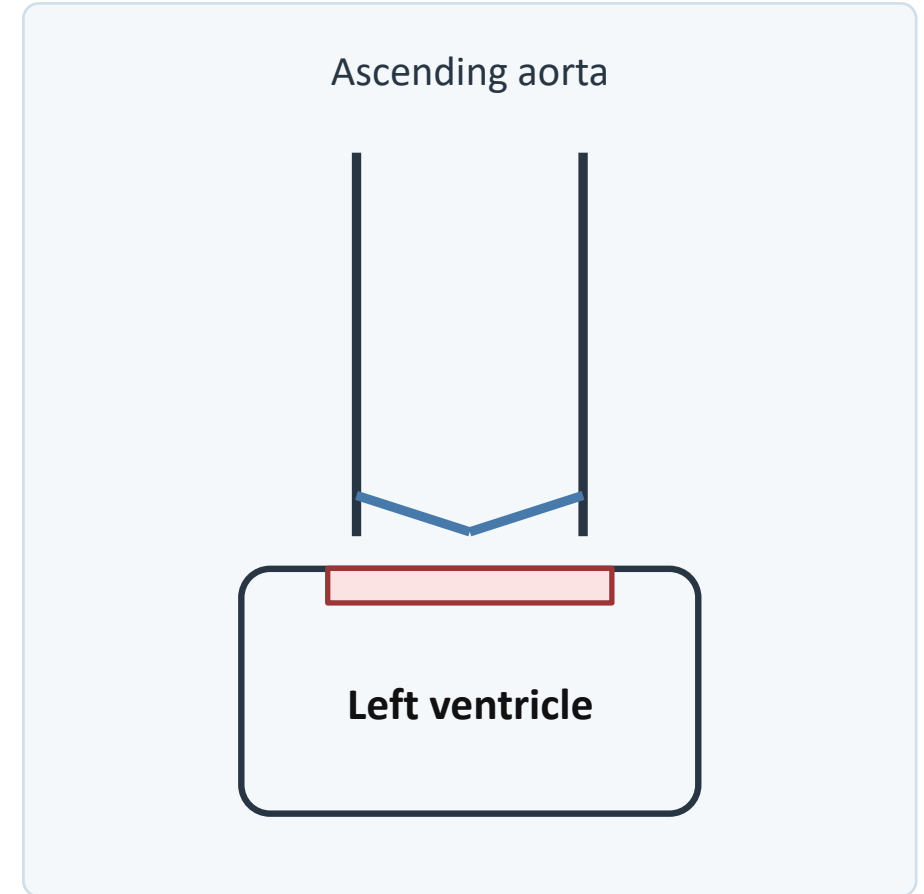
The narrowing is in the left ventricular outflow tract proximal to the valve.



- The aortic cusps sit above the obstructing lesion.
- The stenotic segment is not a malformed ascending aortic wall.
- Localization is essential for morphologic classification.

The subaortic lesion is fibrous tissue

- A thickened ring or collar beneath the aortic cusps.
- The ring consists of dense endocardial fibrous tissue.
- Its encroachment on the outflow passage creates the stenotic lesion.
- The named pathologic structure is a discrete subvalvular collar.



A fibrous collar and cusp dysplasia differ

Subaortic fibrous ring

- Located below the cusps.
- Made of dense endocardial fibrous tissue.
- Constrains the LV outflow passage.

Valvular dysplasia

- Located within the aortic cusps.
- Cusps are thickened and nodular.
- Restricts valvular opening.

Subaortic obstruction has examination clues

- A prominent systolic murmur is typical description.
- A palpable thrill may accompany the murmur.
- These clues indicate obstructed ejection but do not substitute for anatomical localization.
- The underlying lesion remains a fibrous subvalvular collar.

Anatomic reasoning

Systolic ejection



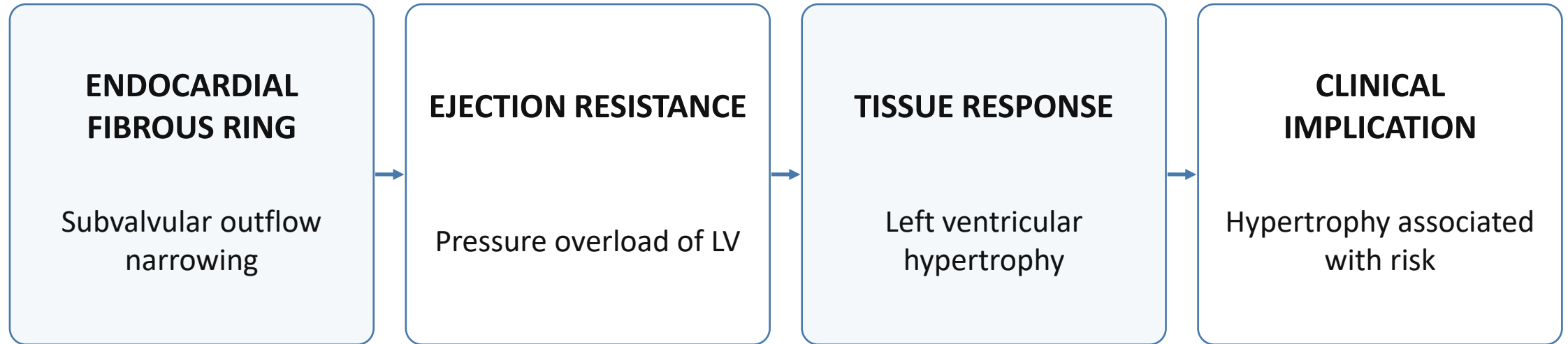
Disturbed flow



Murmur ± thrill

Schematic

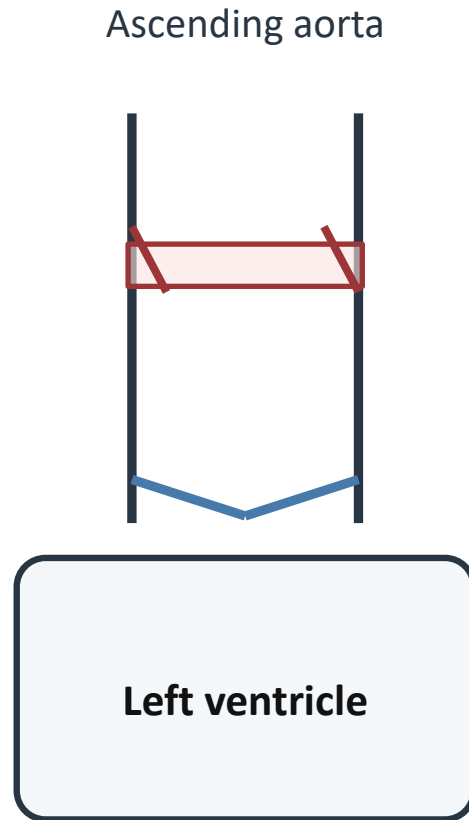
Subaortic obstruction produces a pressure response



Causal sequence / anatomic reasoning schematic

Supravalvular stenosis lies in the aortic wall

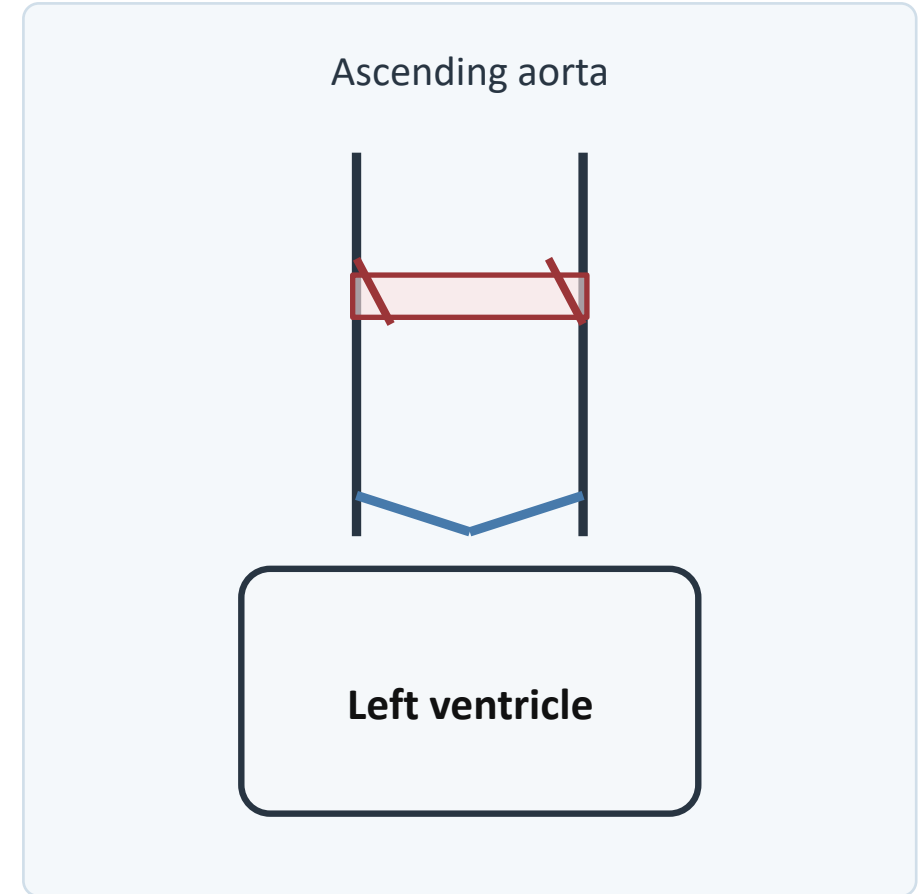
The primary abnormality is superior to the aortic valve.



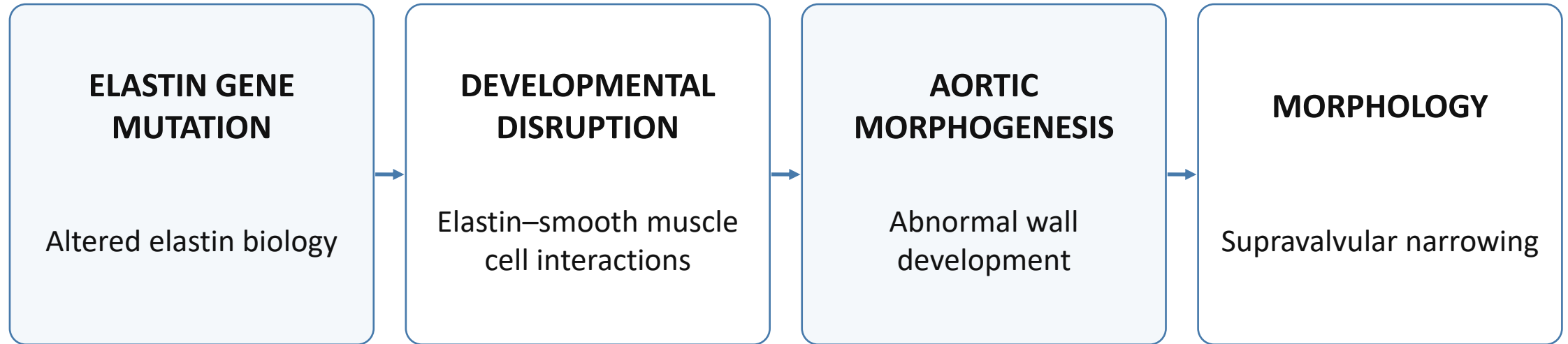
- The ascending aortic wall is thickened and constricted.
- The condition as congenital aortic dysplasia.
- The obstruction is arterial, rather than a malformed cusp or subvalvular membrane.

Supravalvular stenosis: Vessel wall pathology

- Congenital dysplasia affects the ascending aorta.
- Wall thickening reduces the available luminal caliber.
- Constriction above the valve impedes left ventricular ejection.
- The structural target is the aortic wall itself.



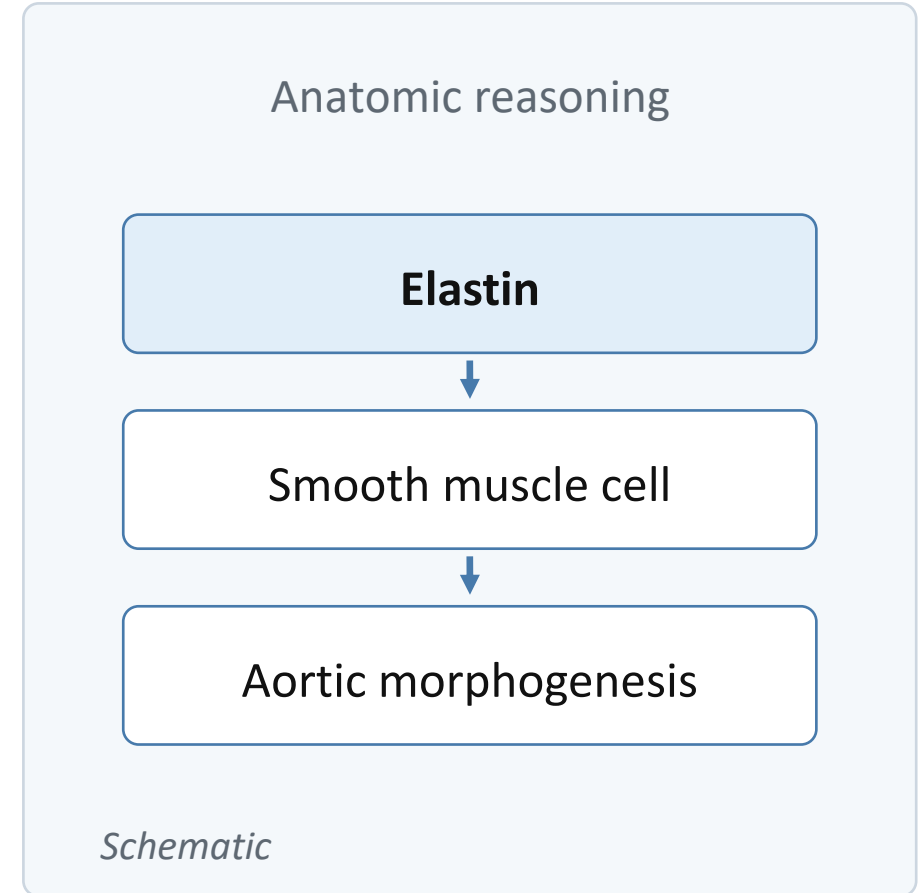
Elastin mutations can cause supravulvular disease



Causal sequence / anatomic reasoning schematic

Elastin–smooth muscle interactions matter

- Elastin is more than a passive structural component of the aortic wall.
- Elastin–smooth muscle cell interactions to aortic morphogenesis.
- Disrupted developmental signaling/structural interaction can contribute to constriction.
- This is a supported mechanism for some supralvalvular disease, not a universal cause of all aortic obstruction.



Genotype and lesion level must be separated

Supported specific association

- Elastin gene mutation.
- Supravalvular aortic stenosis.
- Abnormal aortic wall development.

Not established by this excerpt

- The same cause for all valvular stenosis.
- A single genetic explanation for subaortic collars.
- A genetic result sufficient to grade clinical severity.

Compare the three structural lesions

Level	Primary abnormality	Main structure affected
Subvalvular	Dense endocardial fibrous collar	LV outflow below cusps
Valvular	Small/dysplastic cusps; commissures abnormal	Aortic valve
Supravalvular	Dysplastic, thickened, constricted wall	Ascending aorta

The same pressure response may have different causes

Anatomic cause	Obstruction located at	LV effect
Cusp malformation	Valve	Pressure hypertrophy may develop
Fibrous collar	Below valve	Pressure hypertrophy may develop
Aortic wall narrowing	Above valve	Outflow load is increased

Congenital aortic stenosis versus coarctation

Aortic stenosis / atresia

- Obstruction at, below, or just above the aortic valve.
- May produce left ventricular hypertrophy.
- Severe valve atresia may be associated with HLHS.

Coarctation of the aorta

- Discrete or tubular narrowing of the aorta near the ductal region.
- May produce proximal hypertension and collateral flow.
- A different great-vessel location and disease unit.

Aortic and pulmonary outflow atresia contrast

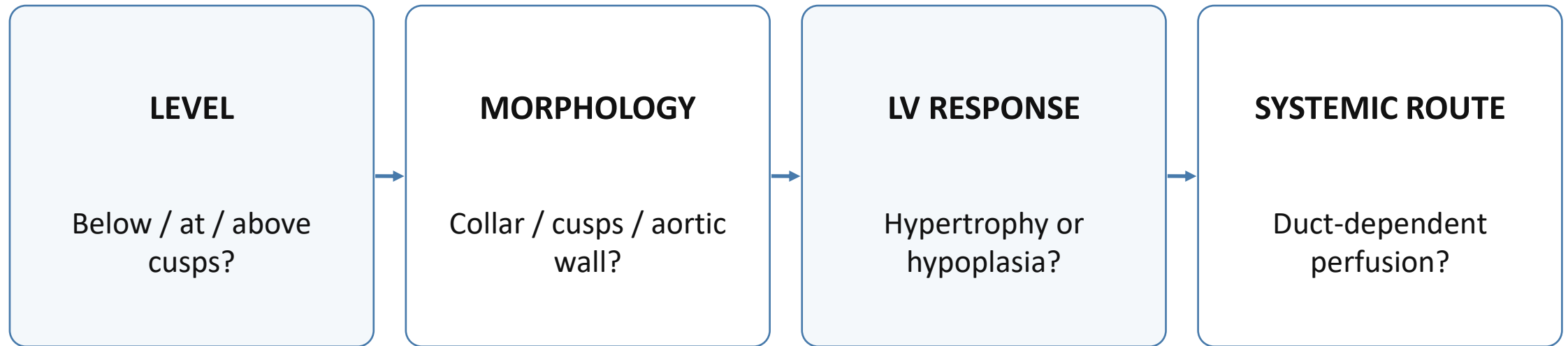
Aortic atresia

- Obstructs left-sided systemic outflow.
- Severe disease may have hypoplastic left ventricle.
- Patent ductus needed for aortic/coronary supply.

Pulmonary atresia

- Obstructs right-sided pulmonary outflow.
- May have hypoplastic right ventricle.
- Patent ductus can provide pulmonary blood supply.

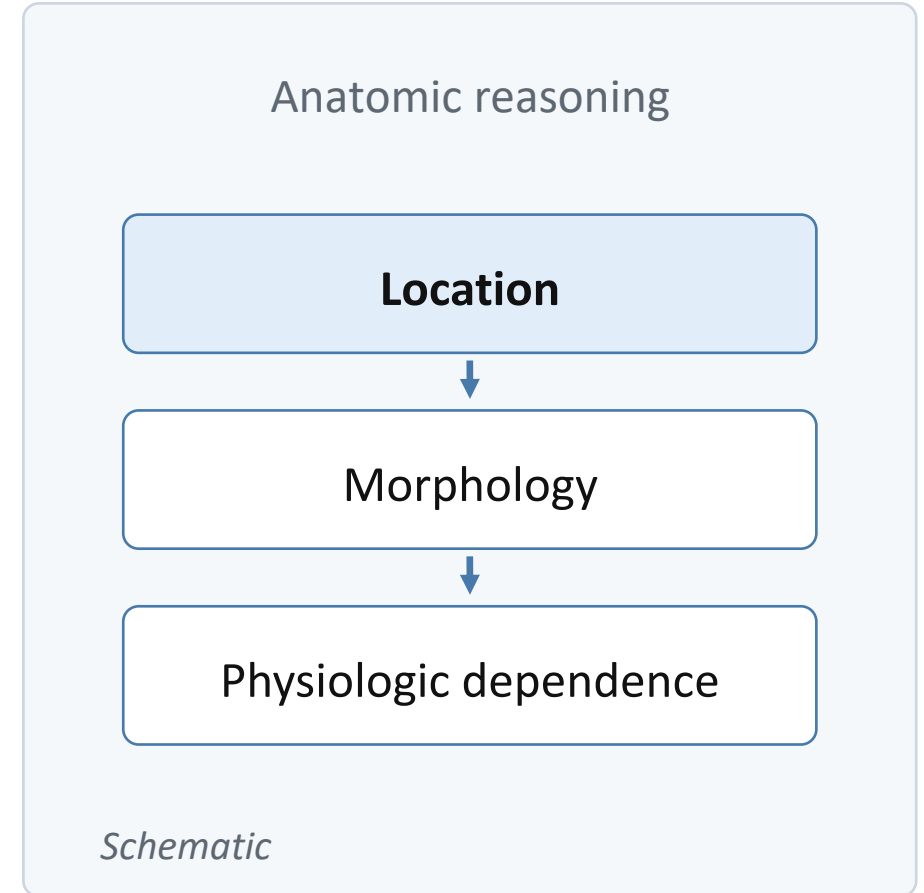
Four structural questions organize evaluation



Causal sequence / anatomic reasoning schematic

Do not confuse clinical severity with anatomy

- Lesion level determines which tissue must be characterized.
- Severity determines how much flow is obstructed and whether compensation is adequate.
- Developmental timing determines whether the ventricle is hypoplastic rather than merely hypertrophied.
- Duct dependence is a circulatory condition, not a synonym for all congenital stenosis.



Morphology-to-clinical correlation

Observation	Pathologic interpretation	Consequence
Small or nodular cusps	Valvular anomaly	Cusp-level narrowing
Fibrous subaortic collar	Endocardial lesion	Subvalvular narrowing
Ascending aortic thickening	Congenital aortic dysplasia	Supravalvular narrowing
Hypoplastic LV + aorta	Severe developmental obstruction	Consider HLHS and duct dependence

Case application: An isolated valve lesion

Hypothetical teaching scenario

Teaching scenario: A child is found to have congenital abnormal aortic cusps. The left ventricle and ascending aorta are adequately developed, and there is no suggestion of duct-dependent systemic flow.

Apply the pathology:

What information supports an isolated valvular lesion rather than hypoplastic left heart syndrome?

Case reasoning: Isolated does not mean harmless

Preserved left-heart growth argues against the severe hypoplastic constellation.

- An abnormal valve can be an isolated lesion.
- A small residual opening can still produce ventricular pressure load.
- The presence of LV hypertrophy does not itself establish HLHS.

Case application: Duct-dependent newborn

Hypothetical teaching scenario

Teaching scenario: A newborn has severe aortic valve obstruction, a diminutive left ventricle and ascending aorta, and systemic blood flow reliant on an open ductus arteriosus.

Apply the pathology:

Which anatomic constellation is present, and why is duct closure critical?

Case reasoning: A duct-dependent syndrome

This matches the severe hypoplastic left heart constellation.

- Aortic stenosis/atresia coexists with LV and ascending aortic hypoplasia.
- The PDA supplies a route for systemic and coronary perfusion.
- Closure in the first week is generally lethal unless patency is preserved.

Case application: A lesion above the cusps

Hypothetical teaching scenario

Teaching scenario: The valve cusps are not the primary lesion. Instead, congenital thickening and constriction affect the ascending aorta above the valve.

Apply the pathology:

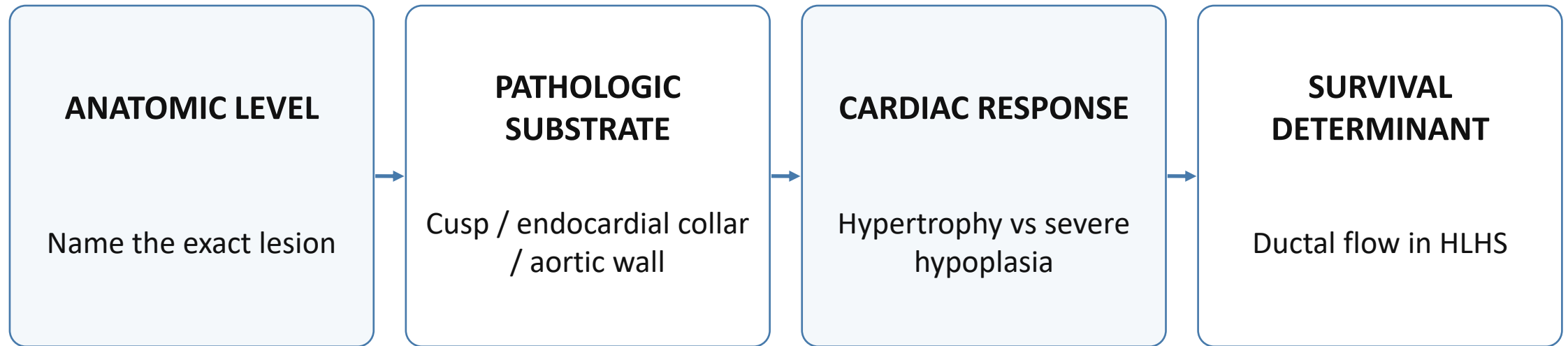
Which category best localizes the defect, and what molecular association does Robbins describe?

Case reasoning: Supravalvular dysplasia

This is supravalvular aortic stenosis.

- The pathologic tissue is the ascending aortic wall.
- Elastin gene mutations can disrupt elastin–smooth muscle interactions.
- The resulting abnormal aortic morphogenesis can produce supravalvular constriction.

Integrated diagnostic synthesis



Causal sequence / anatomic reasoning schematic

Essential pathology conclusions

- Congenital obstruction occurs at three anatomical levels with different pathologic substrates.
- Valvular abnormalities include small or dysplastic cusps and altered commissures.
- Proximal pressure overload explains left ventricular hypertrophy and its potential risks.
- Severe atresia with LV/aortic hypoplasia is duct-dependent and potentially lethal after duct closure.

SUPRAVALVULAR

Ascending aorta

VALVULAR

Valve cusps

SUBVALVULAR

Subaortic collar

Primary reference

Robbins, Cotran & Kumar. Pathologic Basis of Disease. 11th edition (2026).

Chapter: The Heart — sections “Congenital Heart Disease,” “Obstructive Lesions,” and “Aortic Stenosis and Atresia.”

Hoffman JI, Kaplan S. The incidence of congenital heart disease. J Am Coll Cardiol. 2002;39(12):1890–1900.