Tetralogy of Fallot with Pulmonary Stenosis

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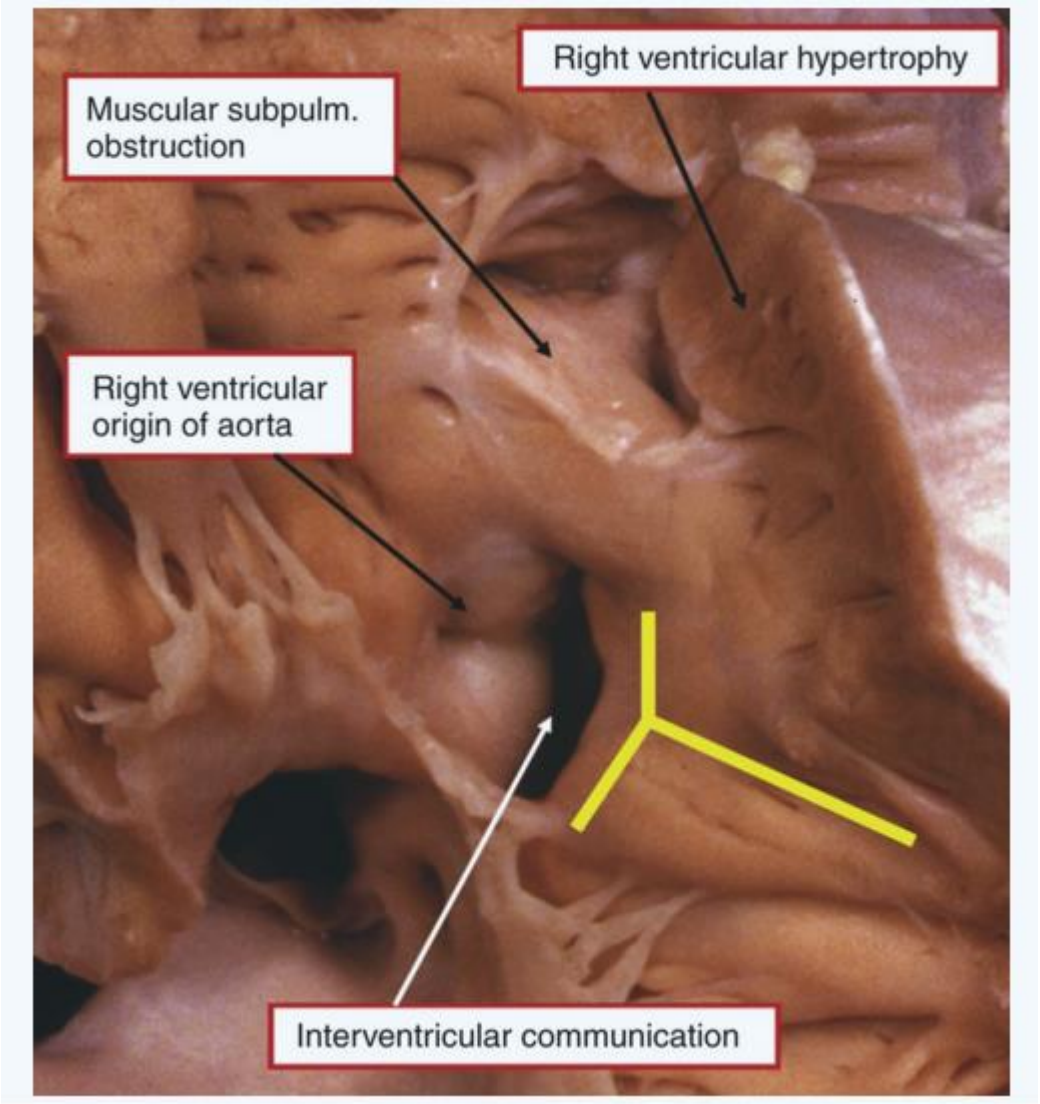
TOF with PS

- Tetralogy of Fallot
- Interventricular communication
- Biventricular origin of the aorta
- Muscular obstruction within the right ventricular outflow tract
- Right ventricular hypertrophy

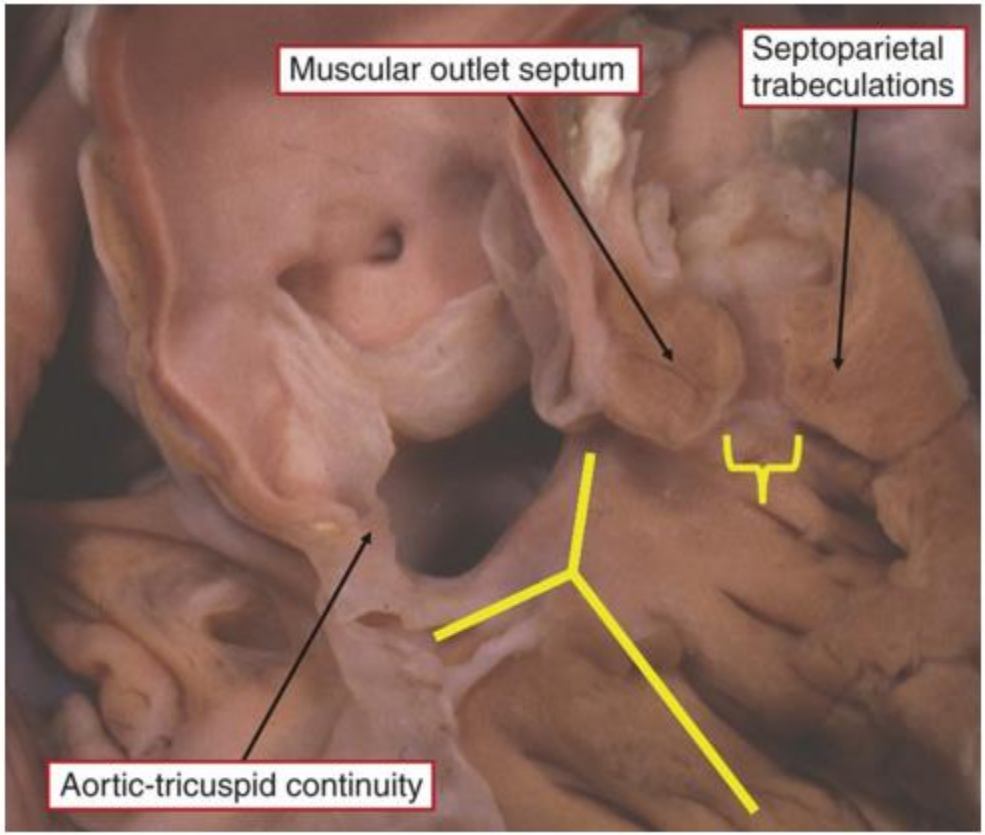


Incidence, prevalence, and aetiology

- 3.5%, of infants born with congenital heart diseass
- 0.28 per 1,000, or 1 in 3,600, live births
- Males and females equally affected
- The risk of recurrence in siblings, about 3% if there are no other affected first-degree relatives
- Microdeletions of the q11 region of chromosome 22, Di George syndrome and the velocardiofacial syndrome, conotruncal anomaly face syndrome
- No differences are found in the incidence of affected children according to whether it is the mother of father who had the lesion initially
- However, the risk is much higher, at above two-fifths, if the affected parent has a sibling with the same or a similar cardiac anomaly

Anatomy and morphogenesis

- Phenotypic features
- Antero-cephalad deviation of the insertion of the muscular outlet septum relative to the limbs of the septomarginal trabeculation, coupled with an arrangement of the septoparietal trabeculations which produces a squeeze at the mouth of the infundibulum

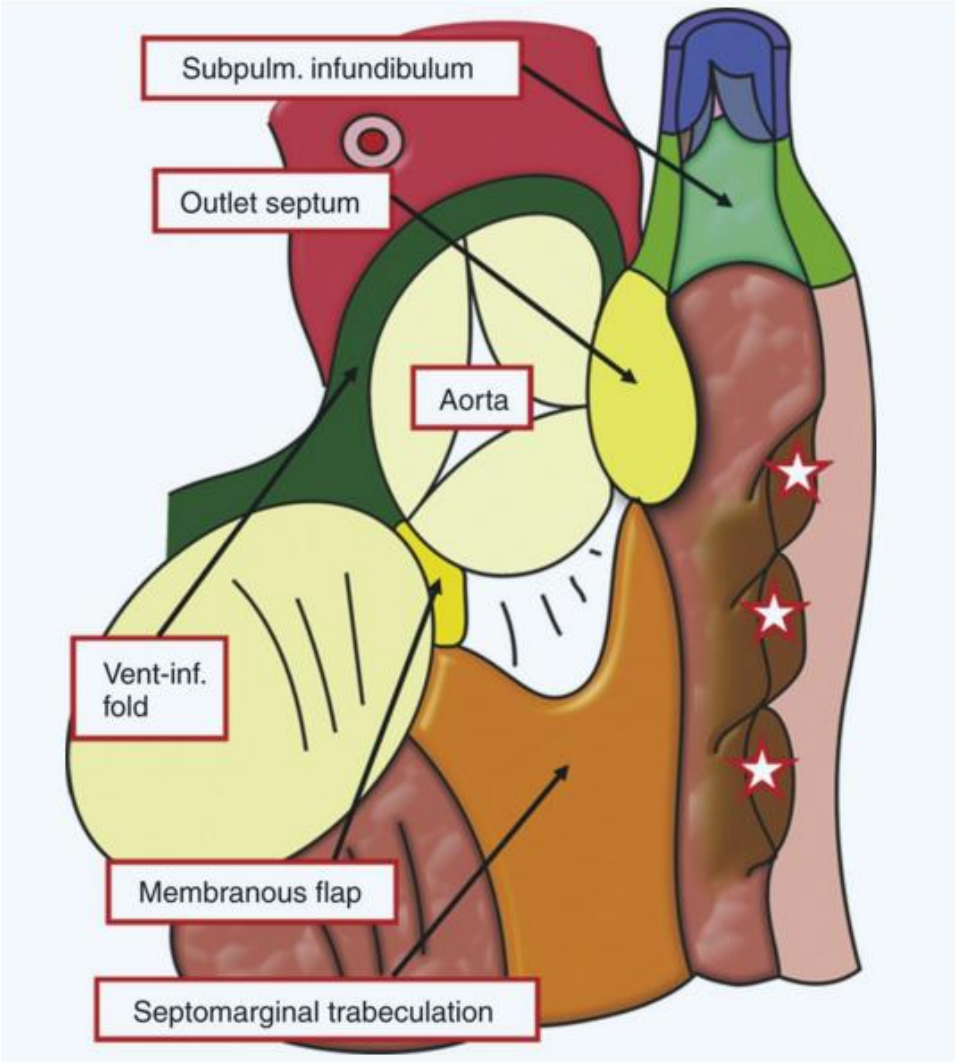


Anatomy and morphogenesis

- Phenotypic features
- The outlet septum can be markedly deviated in antero-cephalad direction without there being subpulmonary stenosis, despite the presence of hypertrophied septoparietal trabeculations, so-called Eisenmenger complex ventricular septal defect

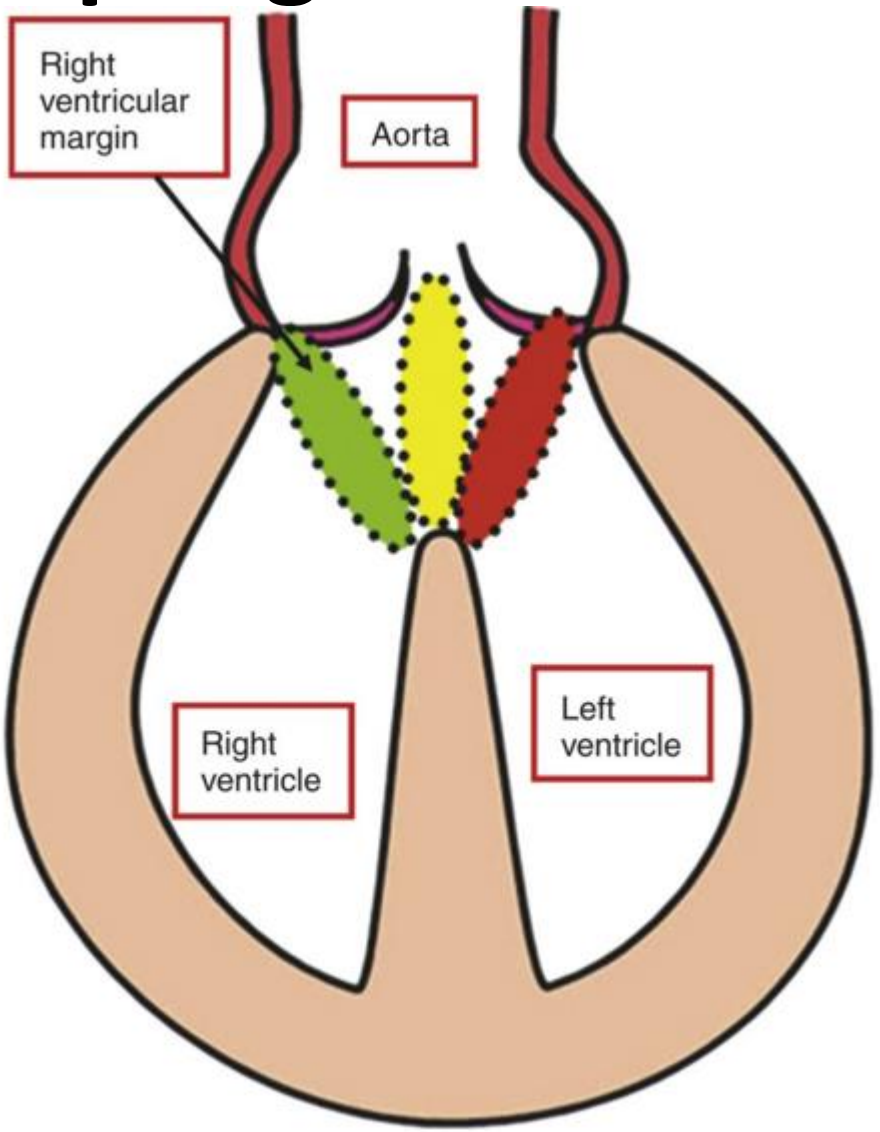
Anatomy and morphogenesis

- Phenotypic features
- Variability in the margins of the ventricular septal defect
- Directly beneath the overriding aortic valve orifice, outlet defect
- Antero-cephalad limb of the septomarginal trabeculation, which forms the anterior margins of the defect
- The roof of the defect is formed by the attachments of the leaflets of the overriding aortic valve to the ventriculo-infundibular fold



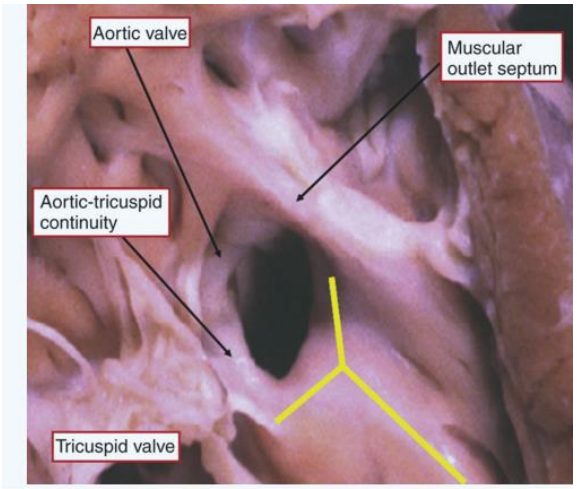
Anatomy and morphogenesis

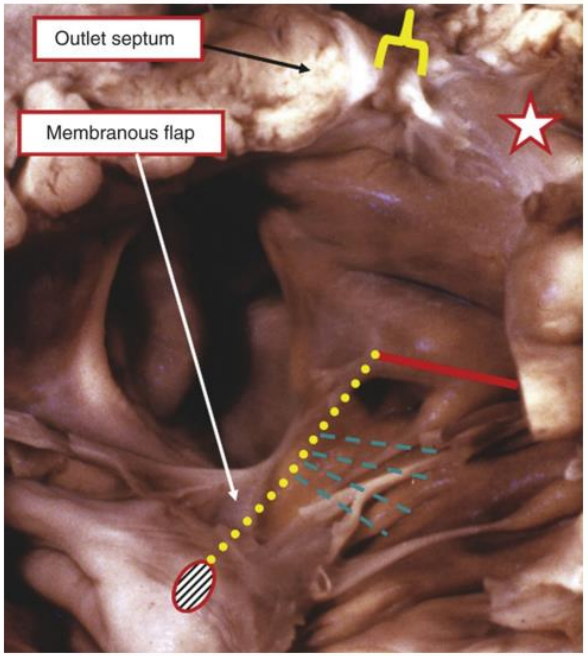
- Phenotypic features
- Variability in the margins of the ventricular septal defect



Anatomy and morphogenesis

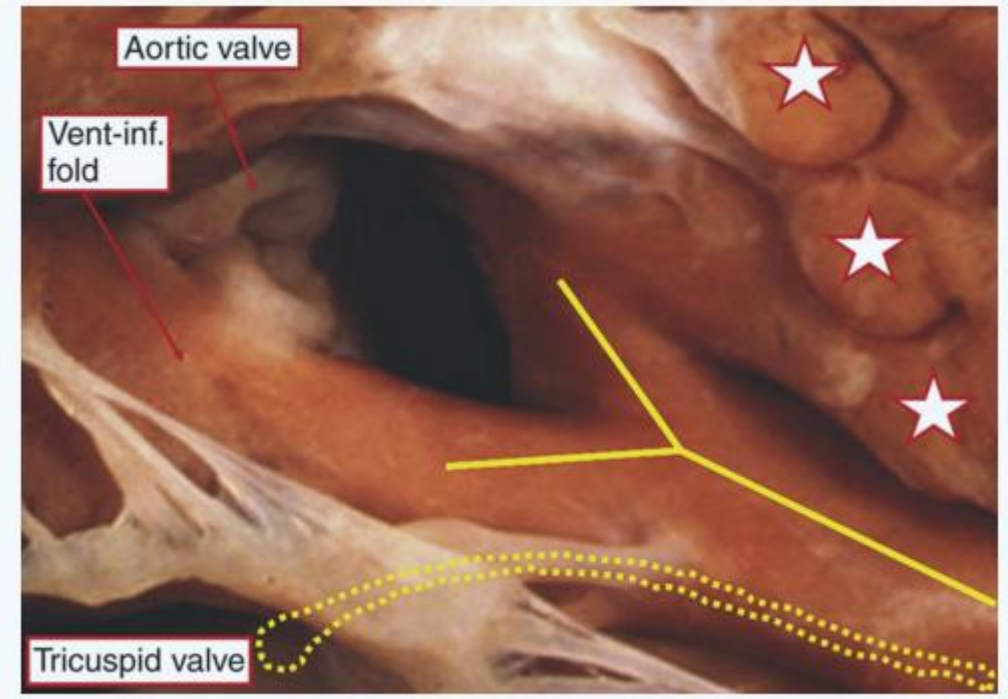
- Phenotypic features
- Variability in the margins of the ventricular septal defect
- In that the postero-inferior margin is an area of fibrous continuity between the leaflets of the aortic and tricuspid valves, perimembranous
- The septal remnant itself, called the membranous flap, is safe tissue for anchorage of sutures when such stitches are placed with care





Anatomy and morphogenesis

- Phenotypic features
- Variability in the margins of the ventricular septal defect
- Second most common pattern, interruption of the area of fibrous continuity between the aortic and tricuspid valves by a muscular fold, muscular outlet



Anatomy and morphogenesis

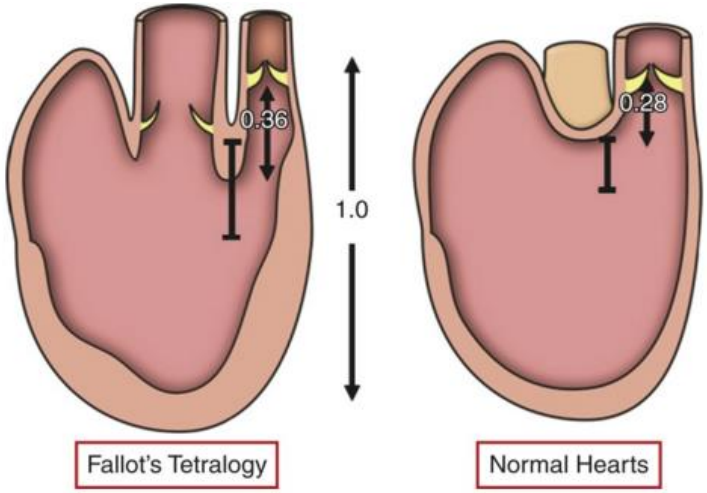
- Phenotypic features
- Variability in the margins of the ventricular septal defect
- The doubly committed and juxta-arterial defect, by far the least common in the Western World, but commoner in the Far East and South America
- Both subaortic and subpulmonary as a consequence of failure of formation of the muscular subpulmonary infundibulum

Anatomy and morphogenesis

- Phenotypic features
- Variability in the margins of the ventricular septal defect
- Inlet defects, associated with straddling and overriding of the tricuspid valve, or the ventricular component of an atrioventricular septal defect associated with common atrioventricular junction

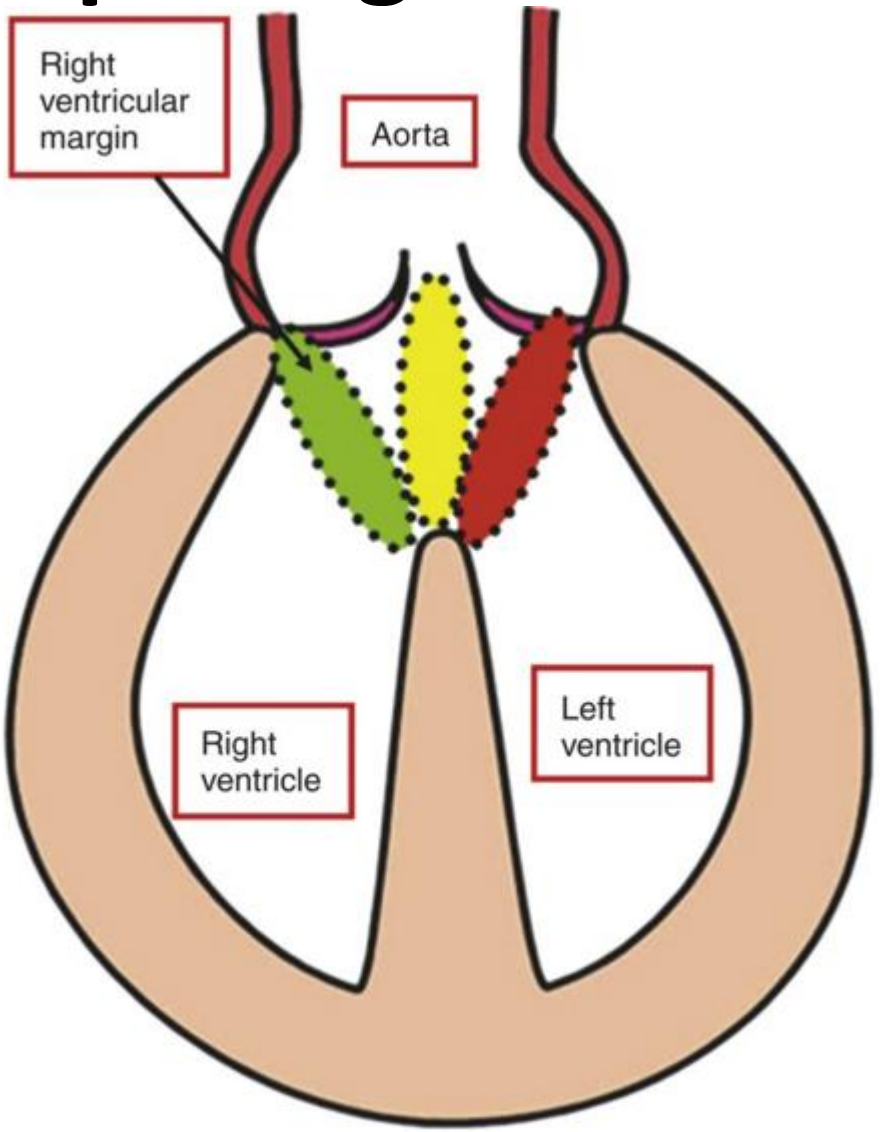
Anatomy and morphogenesis

- Phenotypic features
- Narrowing of the subpulmonary infundibulum
- Subpulmonary stenosis, an essential part of tetralogy, due to the squeeze between the antero-cephalad deviation of the outlet septum and the abnormal arrangement of the distal septoparietal trabeculations
- The infundibulum is longer in the setting of the malformation
- Obstruction at valvar level, two leaflets
- Pulmonary arterial stenosis



Anatomy and morphologenesis

- Phenotypic features
- Overriding of the aortic valve
- Much larger patch will be required, the larger part of the VSD circumference is supported by the right ventricle



Anatomy and morphogenesis

- Phenotypic features
- Other lesions of the pulmonary circulation
- Pulmonary valvar stenosis, stenosis of a bicuspid valve or to stenosis of a valve with three leaflets
- Absence of the leaflets of the pulmonary valve (absent pulmonary valve syndrome), with dilation of the pulmonary trunk and its branches -> airway problems
- Left pulmonary artery connected by the arterial duct
- Right pulmonary artery may arise directly from the ascending aorta
- Major systemic-to-pulmonary collateral arteries are sometimes present, but in association with normal right and left pulmonary arteries

Anatomy and morphogenesis

- Phenotypic features
- Associated anomalies
- ASD
- AVSD, straddling of the tricuspid valve
- Anomalous origin of the anterior interventricular coronary artery from the right coronary artery
- Right aortic arch
- Aortic incompetence, commoner in older patients

Anatomy and morphogenesis

- Morphogenesis
- Study of embryos, in which developmental stages of these malformations were observed, showed that the factory for production of the lesions was within the ventricular outflow tracts
- Deletion of chromosome 22q11 also support the existence of malseptation of the outflow tracts in humans, and point to this being due to problems in migration of cells from the neural crest

Clinical diagnosis

- Dominated by the degree of muscular obstruction of the right ventricular outflow tract
- Modified by associated anomalies, such as persistent patency of the arterial duct, or presence of large systemic-to-pulmonary collateral arteries

Clinical diagnosis

- Presentation when subpulmonary obstruction is severe from birth
- Presentation in the neonatal period
- Severe arterial desaturation, metabolic acidosis -> increased respiratory rate -> concomitant fall in arterial content of carbon dioxide -> compensatory respiratory alkalosis
- Cyanosis, dominant clinical picture, increases with crying, feeding, other activities
- Sometimes, pulmonary circulation is duct-dependent -> maintenance of ductal patency, usually by infusion of prostaglandin E

Clinical diagnosis

- Presentation when subpulmonary obstruction is moderate at birth
- Acyanotic at birth
- Development of cyanosis, dependent on increasing infundibular stenosis, and not on the degree of aortic override

Clinical diagnosis

- Presentation when subpulmonary obstruction is minimal at birth
- Similar to those of a large ventricular septal defect
- Increasing right ventricular hypertrophy -> subpulmonary obstruction more marked -> shunt is reversed -> exhibit the signs and progression as the group with moderate obstruction

Clinical diagnosis

- Presentation with absent pulmonary valve
- Absence of the leaflets of the pulmonary valve
- Respiratory symptoms of inspiratory and expiratory stridor, dyspnea caused by lobar collapse or, at times, lobar emphysema
- Compression of the bronchial tree by the grossly dilated proximal pulmonary arteries

Clinical diagnosis

- Squatting
- Causes an abrupt increase in systemic venous return and a rise in systemic vascular resistance -> right-to-left shunt decresed
- Hypercyanotic attacks
- Lead to reduced cardiac output, and be accompanied by transient loss of consciousness
- Most common between 6 months and 2 years of age

Clinial diagnosis

- Hypercyanotic attacks
- Initial presentation, a history of episodic loss of consciousness, convulsions, episodes of going floppy or pale, transient vacant episodes, episodes of becoming deeply cyanosed followed by loss of consciousness or sleep
- Episodes of very rapid deep respiration or hyperpnoea, or a high-pitched abnormal cry
- Spells results from infundibular spasm or shutdown

Clinical diagnosis

- Hypercyanotic attacks
- Shutdown, secondary to other primary physiologic influences, such as dehydration, or tachycardia-induced reduction in right ventricular preload, systemic vasodilation in response to fever, or other sympathetic activity
- Prompt treatment with continuous b-blockade, and referral for surgery or interventional catheterization

Clinical diagnosis

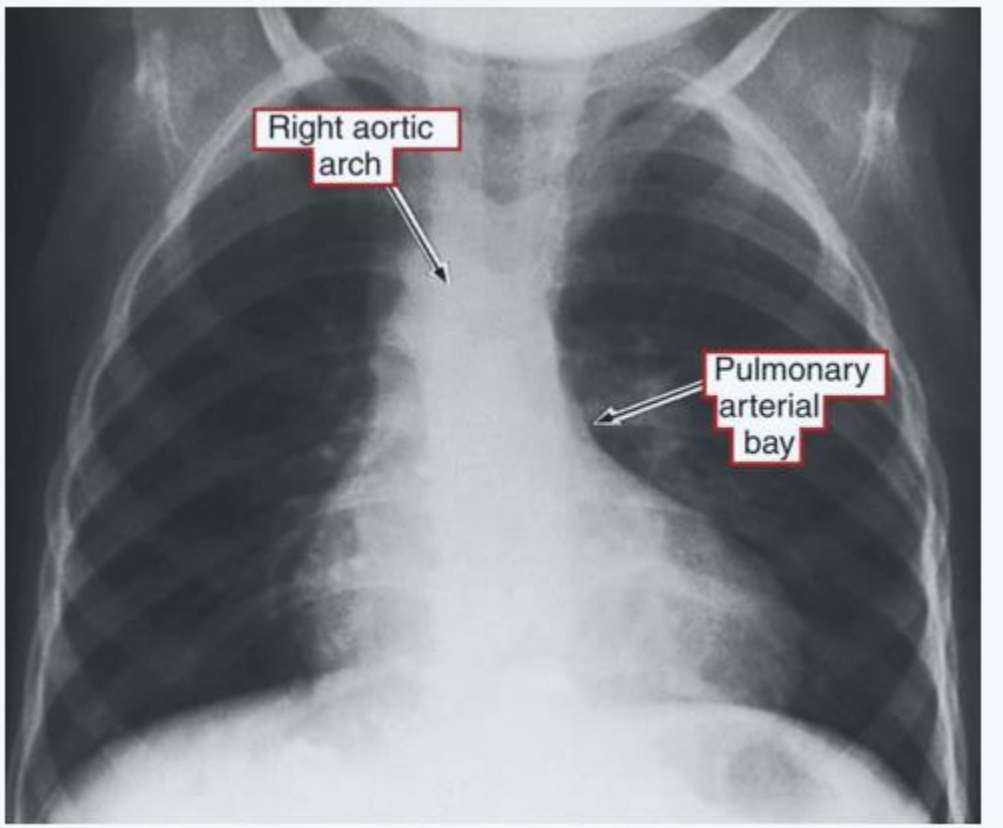
- Physical examination
- Cyanosis
- Auscultation, a systolic ejection murmur with a single second heart sound
- Overt clubbing of fingers and toes, not detected until 2 or 3 months of age
- Some degree of facial dysmorphism, associated syndromes, DiGeorge, Goldenhauer, or Down syndromes
- All patients should now undergo chromosomal analysis (22q11 deletion)
- Loud continuous murmur, more likely to originate from flow through large major systemic-to-pulmonary collateral arteries than the arterial duct

Clinical diagnosis

- Physical examination
- Subpulmonary obstruction, minimal or absent, tachpnoea, dyspnea, and intercostal or subcostal recession (heart failure symptoms)

Clinical diagnosis

- Investigations
- Confirmation is now provided largely by cross sectional echocardiography
- Chest radiograph
- Up to one-third of patients with tetralogy right aortic arch
- Reduced pulmonary vascular markings
- Pulmonary bay
- The apex of the heart may be upturned, because the hypertrophied right ventricle forms the apex in the postero-anterior projection

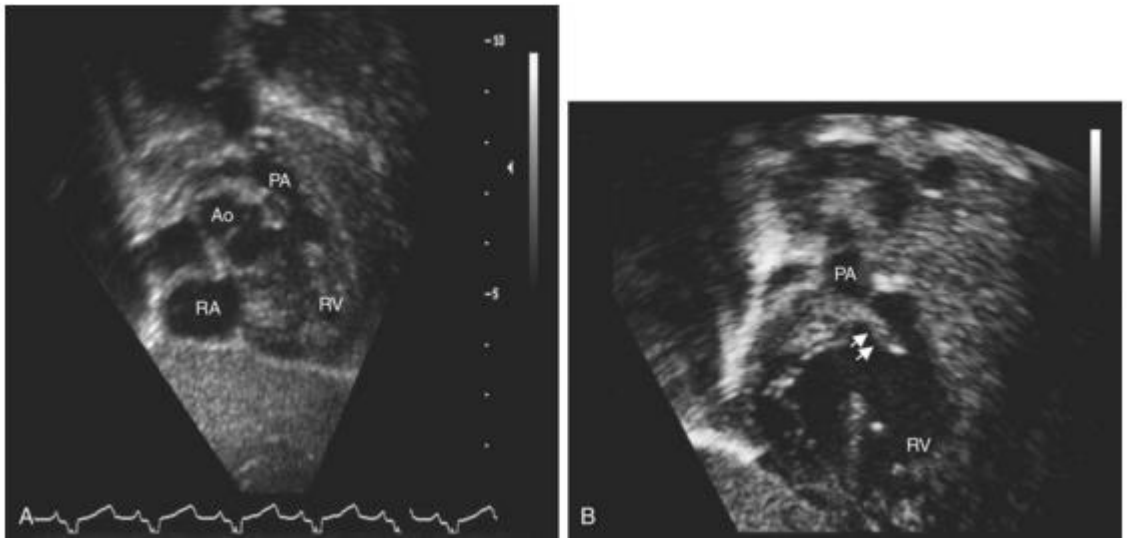


Clinical diagnosis

- Investigations
- Electrocardiography
- Normal or rightward QRS axis and overt right ventricular hypertrophy
- After surgery, right bundle branch block with prolongation of the QRS duration is frequent

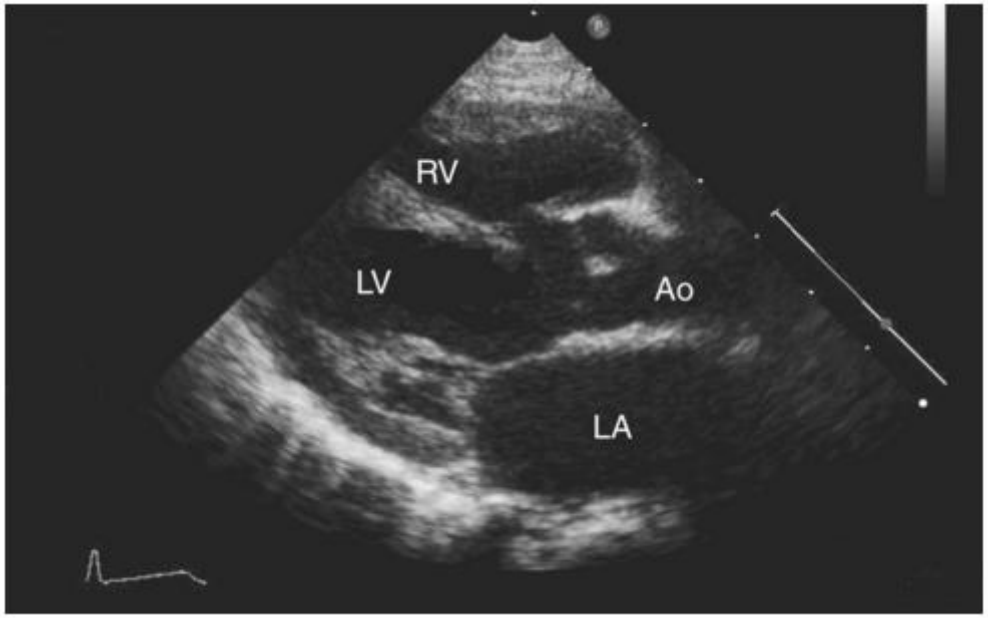
Clinical diagnosis

- Investigations
- Echocardiography
- Subcostal paracoronal view, narrowed subpulmonary outflow tract, with malalignment of the anteriorly displaced muscular outlet septum



Clinical diagnosis

- Investigations
- Echocardiography
- Parasternal long-axis views, aortic override



Clinical diagnosis

- Investigations
- Echocardiography
- Four-chamber view, straddling or overriding tricuspid valve, common atrioventricular junction in hearts with deficient atrioventricular septation
- When the pulmonary trunk is traced to its bifurcation, it is possible to determine the size of the pulmonary arteries to the level of their first bifurcation, but not the more distal pattern of branching
- To confirm normal systemic and pulmonary venous connections
- Anomalous arteries can be identified crossing the subpulmonary infundibulum (coronary arterial anatomy)

Clinical diagnosis

- Investigations
- Echocardiography
- Post-operative assessment
- Presence of residual lesions
- Pulmonary regurgitation
- Right ventricular performance

Clinical diagnosis

- Investigations
- Magnetic resonance imaging
- Post-operative evaluation and decision-making
- Quantitative assessment of right ventricular volume, mass, and function
- Imaging of delayed enhancement using gadolinium, any scarring or fibrosis of the ventricles, contributing to an increased propensity for arrhythmias, or formation of aneurysms in the right ventricular outflow tract

Clinical diagnosis

- Investigations
- Cardiac catheterization and angiography
- To assess the haemodynamics within, and connections between, the individual vessels
- Dilation and stenting of the right ventricular outflow tract
- Pre-operative balloon dilation of the pulmonary arteries, stenting of the arterial duct, interventions on the aortopulmonary collateral arteries
- Additional postoperative interventions
- CT angiography

Haemodynamics and physiology

- Dominated by the severity of obstruction within the subpulmonary right ventricular outflow tract
- The relative flows in the pulmonary and systemic circuits depend on the relative resistances, or impedances

Therapeutic options

- Medical management
- In the knee-chest position and administer oxygen by face mask
- Intravenous line, small dose of morphine sulphate, at 0.1mg per kg
- B-blocking agent such as propranolol (reduce tachycardia and increase systemic resistance, reduce hypercontracility)
- Accompanying metabolic acidosis should be corrected
- Intubation and ventilation in extreme cases, intravenous vasoconstrictor, phenylephrine
- Emergency systemic-to-pulmonary shunt

Therapeutic options

- Surgical management
- Those over a few months of age, definite surgery
- Neonates and young infant, surgical palliation by construction of a systemic-to-pulmonary arterial shunt, balloon dilation and stenting of the right ventricular outflow tract, and stenting of the arterial duct

Therapeutic options

- Surgical management
- Palliative procedures
- Modified BT shunt, now frequently performed through a median sternotomy
- Heparin in the initial post-operative period, followed by aspirin until the time of corrective surgery
- Stenosis of the PA, potential damage to the recurrent laryngeal or phrenic nerves, and adverse physiologic effects such as increased flow of blood to the lungs, systemic steal syndrome

Therapeutic options

- Surgical management
- Definitive repair
- Rarely performed via a large ventriculotomy, with tranatrial closure of the septal defect now almost universal
- Hypertrophied outlet septum, together with its parietal and septal extensions, is excised (infundibulectomy)
- Transannular patch repair vs. pulmonary annulus saving or small annulotomy
- MVOP (monocusp valved outflow patch)
- Ideal surgery, normal right ventricular pressures, absence of any gradient in the right ventricular outflow tract -> to preserve the pulmonary valve, even at the expense of a modest degree of residual stenosis (minimize the adverse late effects of pulmonary incompetence, and retain the integrity of the outflow tract, avoiding late dilation and formation of aneurysms)

Therapeutic options

- Surgical management
- Definitive repair
- The timing of definitive repair, based on the results in the institution offering treatment, usually between 3 and 6 months of age

Therapeutic options

- Surgical management
- Early outcomes
- First feature, diffusely small pulmonary arteries, the ratio of the combined diameter of the right and left PAs, measured just before of their first bifurcation, to that of the descending aorta is less than 1.5 to 1, not attempting primary repair
- Second factor, PAs abnormalities, anomalous origin of one artery, or stenosis of either artery at its origin
- Third factor, small pulmonary valvar orifice (small annulus)
- Final factor, age at repair, both old and young, more than 5 years

Therapeutic options

- Surgical management
- Early post-operative complications
- Post-operative low cardiac output, inadequate relief of subpulmonary obstruction, or an obstructed or restrictive pulmonary vascular bed
- Post-operative pulmonary incompetence
- Consists of maintenance of sinus rhythm and right ventricular preload, keeping the central venous pressure at 12 to 15mmHg, early drainage of resulting effusions, and early extubation
- Early post-operative restriction is a transient phenomenon, usually resolving within 72 hours, although reappearance in the later postoperative follow-up period

Therapeutic options

- Surgical management
- Late outcomes of intervention
- The adverse affects of pulmonary incompetence, previously considered to be a benign side-effect of relief of the obstructed right ventricular outflow tract

Therapeutic options

- Surgical management
- Physical response to correction
- Residual ventricular septal defect
- Right bundle branch block, duration of the QRS complex lengthening in response to dilation of the right heart
- Pulmonary incompetence, remain asymptomatic for up to 20 years, but thereafter freedom from symptoms declines
- Right-sided failure, arrhythmias, and sudden death
- Decreased exercise performance, and right ventricular function
- Nowadays, pulmonary incompetence be measured directly and accurately by cardiac magnetic resonance

Therapeutic options

- Surgical management
- Physical response to correction
- In the context of the mechanical dysfunction of the right ventricle, increasingly appreciated that additional left ventricular dysfunction seems to co-exist in many patients with tetralogy -> biventricular resynchronization -> significant increase in cardiac output
- Aortic dilation and valvar incompetence increasingly with duration of follow-up

Therapeutic options

- Surgical management
- Post-operative conduction disturbances and arrhythmias
- Ventricular arrhythmias, rather than conduction defects, are more likely to be the basis for sudden death
- Older age at operation, more extensive surgery, extent of the ventriculotomy are associated with an increase in ventricular arrhythmias
- Sudden death occurs in about 6% of patients over the long term
- QRS prolongation, right ventricular dilation, sustained ventricular arrhythmias, related to late sudden death
- It must also not be forgotten that atrial arrhythmias

Therapeutic options

- Surgical management
- Treatment of arrhythmias and conduction disturbances at follow-up
- The aims of treatment are relief of symptoms and prevention of sudden death
- Routine treatment of asymptomatic patient with non-sustained tachycardia is not currently indicated
- Implantable defibrillator
- Atrial arrhythmias -> medical therapies, catheter-based ablation, additional maze procedure

Therapeutic options

- Surgical management
- Treatment of pulmonary valve incompetence
- Surgical replacement of the pulmonary valve in symptomatic patients
- Optimal timing of pulmonary valvar replacement, a threshold of right ventricular end-diastolic volume for intervention, in order to preserve the likelihood of adequate reverse remodeling, is in the resion of 170 to 200 mL per square meter of body surface area
- Transcatheter implantation of the pulmonary valve

Therapeutic options

- Surgical management
- Other reinterventions
- Residual pulmonary stenosis, resection of residual infundibular stenosis, or placement of a patch in the right ventricular outflow tract
- Excision of the aneurysmally dialted patch
- Severe tricuspid insufficiency, valvar annuloplasty
- Aortic valve replacement, or the aortic root replacement (>55mm in diameter)
- Residual VSD closure
- RPA or LPA stenosis, balloon dilation, stent insertion, or surgery

Recommendations for long-term follow-up

- Investigations and frequency
- All patients should have regular follow-up by an appropriately trained cardiologist at least once per year
- Pregnancy
- All patients with tetralogy, cardiologic counseling prior to conception, and follow-up by an appropriately trained cardiologist during pregnancy
- Genetic counselling
- The risk of recurrence in offspring of parents with tetralogy, 2% if the mother affected, 1.4% if the father has tetralogy of Fallot
- 22q11 deletion, the risk of transmission, 50%
- The risk of pregnancy in patients with tetralogy of Fallot after corrective surgery depends on the haemodynamic state
- In patients with significant residual obstruction across the right ventricular outflow tract, severe pulmonary regurgitation, tricuspid regurgitation, and right ventricular dysfunction as well as left ventricular dysfunction, the increased volume load of pregnancy may lead to right heart failure and arrhythmias
- Peripartal antibitotic prophylaxis is recommended