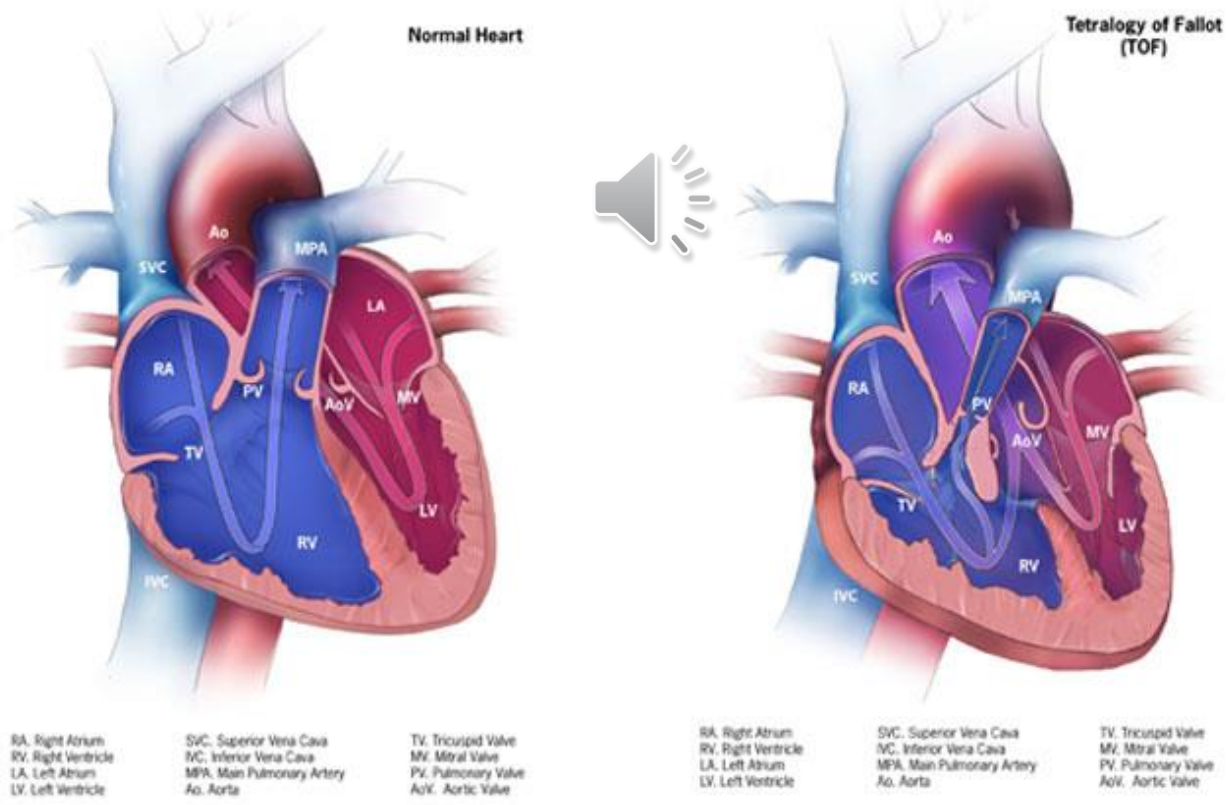
CYANOTIC HEART DEFECTS
TETRALOGY OF FALLOT



ToF makes up 10% of congenital heart defects and is the commonest cyanotic lesion. 25% of patients have DiGeorge syndrome or deletion/ abnormalities of chromosome 22.

• It is usually an isolated defect, and is characterized by four lesions caused by anterior deviation of the outlet septum:

1- Pulmonary stenosis (at the infundibulum, valve or the PA).

2- RV hypertrophy.

3- Overriding aorta.

4- VSD.

• Children present soon after birth with cyanosis, depending on the severity of the pulmonary stenosis, and are usually managed with surgical correction.

PATHOPHYSIOLOGY

The RVOT obstruction *reduces pulmonary blood flow* and also increases the RV pressure causing *a right-to-left shunt* across the large, unrestricted VSD. Both these effects cause *cyanosis*. Immediately after birth, the ductus arteriosus supplies blood to the pulmonary vasculature, but after it closes there may be a sudden onset of cyanosis, and the RV will start to hypertrophy. In a small subset, the RVOT obstruction is so mild as to avoid cyanosis: '*acyanotic*' *or* '*pink*' *tetralogy*. The pulmonary vasculature is protected from the effects of the VSD by the outflow obstruction, and some patients may be well balanced for years.

PRESENTATION AND NATURAL HISTORY

In children with a very hypoplastic annulus (pulmonary stenosis), the presentation is usually soon after birth, with cyanosis due to the obstruction in pulmonary blood flow and thus increased flow across the VSD from right to left. In children with a larger pulmonary valve annulus, presentation is usually later, and may be with 'tet spells'—cyanotic episodes that occur when there is muscular spasm causing acute obstruction to RV outflow and reversal of the shunt(also due to falling SVR with exercise). 'Squatting' is a characteristic compensatory maneuver, seen in right-to-left shunts developing on exercise.

Hemoptysis may be present late. Examination shows RV heave and a systolic ejection murmur. The degree of RVOT obstruction is the main determinant of outcome in untreated ToF: most children develop signs and symptoms by 6 months of age, and <10% survive to the age of 21 without surgical intervention. 25% of untreated patients die within 1 year of life, 40% of untreated patients die within 4 years, and 95% of untreated patients die within 40 years.

INVESTIGATIONS

- EKG shows right-axis deviation, RVH and RA hypertrophy.
- CXR shows a 'boot-shaped' heart as a result of RVH and the small or absent MPA; and decreased pulmonary vascularity.
- Echo is diagnostic, and defines the severity and extent of the RVOT obstruction, the size of the pulmonary valve annulus, the size and position of the VSD, the coronary artery arrangement, and any associated anomalies.
- Cardiac catheterization is used if there is a need to further image the coronary arteries, or if there are concerns about the size of the pulmonary arteries.

SURGICAL MANAGEMENT

There is an increasing trend towards earlier complete repair, but there is still a place for early palliation with a *systemic–PA shunt* followed by repair at a later stage. A sensible approach is to aim to repair all tetralogies between 3–6 months. If they present with cyanotic spells they can be repaired earlier, or if institutional policy dictates, undergo a *B-T shunt* with elective repair when they are bigger.

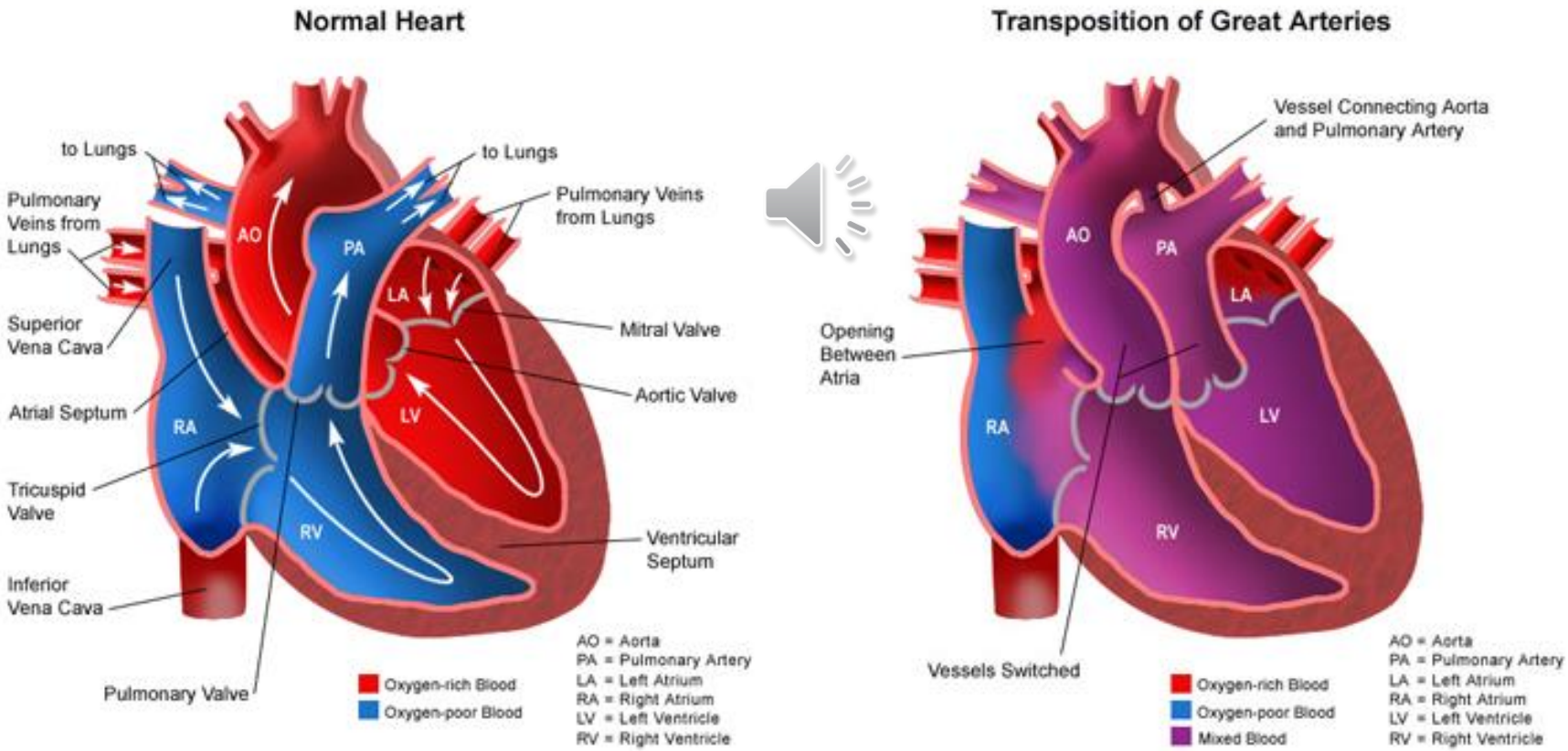
TRANSPOSITION OF GREAT ARTERIES

• TGA is a congenital cardiac anomaly in which the aorta arises from the RV and the pulmonary trunk arises from the LV.

• Complete TGA is the most common cyanotic lesion presenting in the neonatal period, and makes up 5% of congenital cardiac lesions. In 90% it is isolated, with no associated extracardiac malformations.

• Infants present with cyanosis and in the absence of an ASD or VSD infants die within a few hours of closure of the ductus arteriosus.

• A corrective arterial switch procedure can be performed in neonates with low mortality.


Normal Heart
to Lungs
to Lungs
Pulmonary Veins from Lungs
Pulmonary Veins from Lungs
Superior Vena Cava
Atrial Septum
Tricuspid Valve
Inferior Vena Cava
Pulmonary Valve
AO
PA
LA
RA
LV
RV
Mitral Valve
Aortic Valve
Ventricular Septum
Oxygen-rich Blood
Oxygen-poor Blood
AO = Aorta
PA = Pulmonary Artery
LA = Left Atrium
RA = Right Atrium
LV = Left Ventricle
RV = Right Ventricle
Transposition of Great Arteries
Vessel Connecting Aorta and Pulmonary Artery
Opening Between Atria
Vessels Switched
AO
PA
LA
RA
LV
RV
Oxygen-rich Blood
Oxygen-poor Blood
Mixed Blood
AO = Aorta
PA = Pulmonary Artery
LA = Left Atrium
RA = Right Atrium
LV = Left Ventricle
RV = Right Ventricle

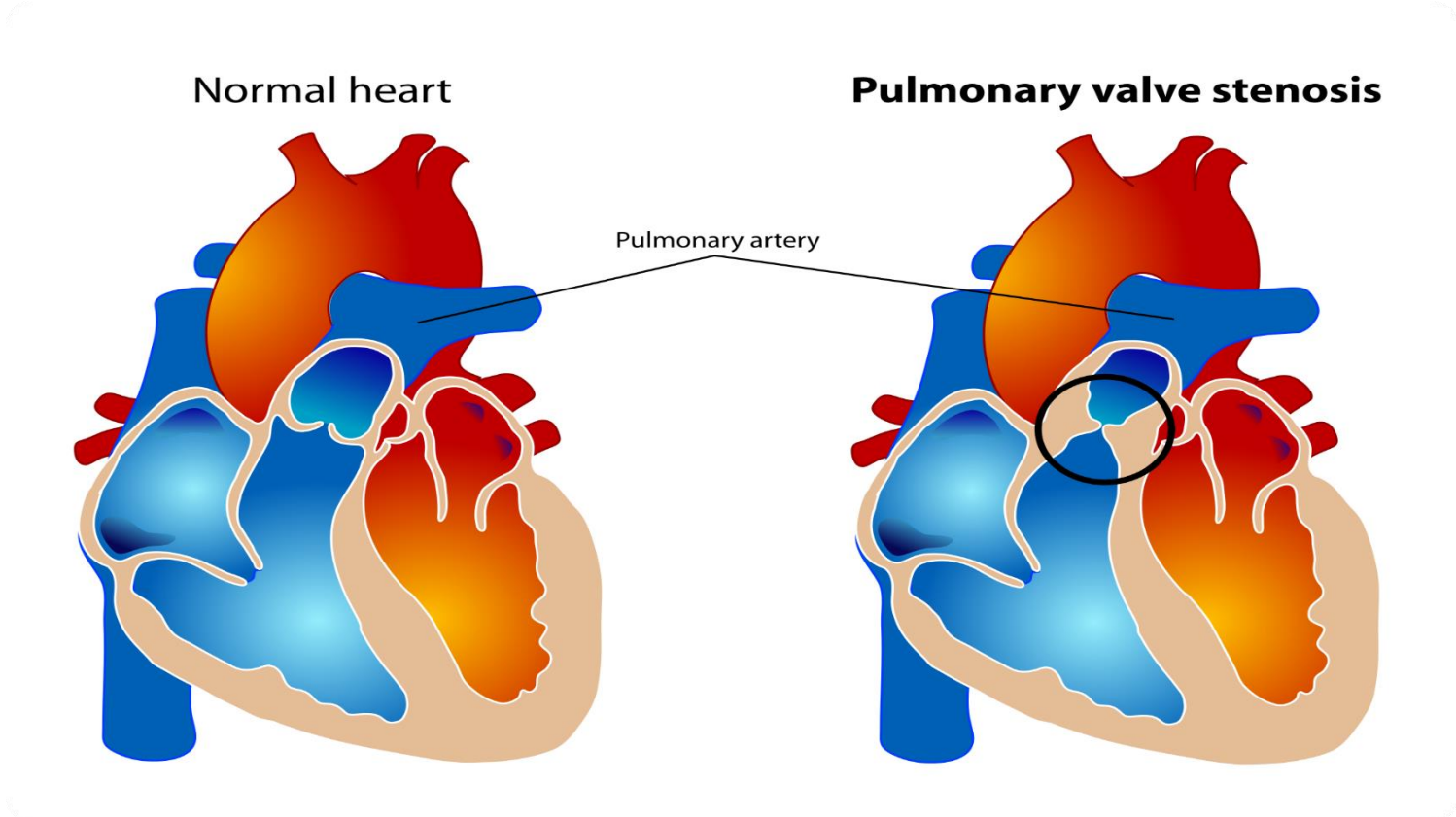
PULMONARY STENOSIS

Key facts

This defect makes up 8–10% of all congenital cardiac defects, and usually involves valvar PS, but includes subvalvar and supravalvar PS.

Morphology

The *pulmonary valve is stenotic*, and usually is tricuspid with fusion of the commissures, thus producing a dome-like structure with a variable central opening. There is often a *PFO or ASD* (40%). There may be *RV hypertrophy*, even at birth, and *subvalvar stenosis* from infundibular muscle hypertrophy. There may be *poststenotic dilatation* of the pulmonary arteries.


Normal heart
Pulmonary valve stenosis
Pulmonary artery

ACQUIRED HEART DISEASES

ISCHEMIC HEART DISEASE

Pathophysiology

• Stenotic coronary artery disease (CAD) is narrowing of the coronary arteries caused by thickening and loss of elasticity of the arterial walls (atherosclerosis).

• Stable plaques are mostly responsible for the lesions seen on angiography and stable angina: MI is due to unstable plaque rupture and thrombosis: 50% of MIs occur distal to angiographically 'normal' vessels.

Symptoms

Exertional angina: this is common. It results from reduction of coronary flow reserve, and the severity depends on the mismatch between myocardial oxygen supply and demand, i.e., both the severity of the CAD and the amount of work the myocardium is required to do.

Dyspnea: graded in the same way as angina A number of mechanisms contribute.

Acutely, transient systolic, and/or diastolic LV dysfunction results from worsening myocardial supply:demand mismatch caused by increases in preload, afterload, hypotension, or exertion

. Orthopnea results from the sudden increase in preload on lying flat. Paroxysmal nocturnal dyspnea is due to pulmonary edema, and poorly understood changes in respiratory drive.

Nausea: parasympathetic stimulation results in nausea and vomiting.

Unstable angina: this term, which covers a multitude of syndromes all of which reflect an adverse prognostic turn, applies to patients with severe and persisting angina. It is most commonly caused by non-occlusive thrombus at the site of an unstable plaque. Unstable angina normally recurs as either another episode of angina or as an acute MI. Patients can be divided into low-, medium-, and high-risk categories depending on the duration of their angina, severity of symptoms, presence of EKG changes, and hemodynamic compromise.

ACUTE CORONARY SYNDROME (ACS)

- This includes unstable angina, NSTEMI, and STEMI.
- Unstable angina is differentiated from NSTEMI by raised cardiac enzymes (CK-MB or troponin) that are present in NSTEMI.
- STEMI is differentiated from NSTEMI by EKG changes.

VALVULAR HEART DISEASE

Aortic stenosis

Etiology

• Congenital (unicuspid rare, *bicuspid valve* more common around 1–2%). Bicuspid valve morphology varies—may be two equal cusps with central opening, or two unequally sized cusps with raphe in larger cusp indicating where two leafl ets have fused. Bicuspid aortic valves usually functionally normal in younger patients, but leaflets may become increasingly sclerotic with age, leading to accelerated stenosis.

Around 50% of bicupisd aortic valve are stenotic by age 60 years.

Bicuspid valves associated with aortic root dilatation .

• *Rheumatic* (b pp372 and 398) due to commissural fusion, leaflet thickening.

• *Calcific degeneration:* commonest cause, occurring in otherwise normal valves. Rheumatic and bicuspid valves eventually calcify.

• Infective endocarditis (rare cause of AS, usually causes AI).

• Hyperlipidaemia (rare).

• Subvalvar (membrane and muscular) and supravalvular.

• Prosthesis failure (pannus, thrombosis, endocarditis, calcification).

Aortic insufficiency

Etiology

• Myxomatous degeneration (common cause, causes leaflet prolapse).

• Rheumatoid (often mixed picture with degree of AS).

• Infective endocarditis (leaflet perforation).

• Root dilatation (quite common—due to rheumatic, atherosclerotic, aneurysmal, Marfan syndrome, syphilis, ankylosing spondylitis).

• Prosthesis failure: paraprosthetic leak, leaflet perforation.

MITRAL STENOSIS

ETIOLOGY

- CHRONIC *RHEUMATIC* HEART DISEASE (COMMONEST CAUSE).
- *CONGENITAL* MITRAL STENOSIS (RARE).
- *MITRAL ANNULAR CALCIFICATION* (MAC) (LESS USUAL CAUSE OF SEVERE MS).

Mitral regurgitation

Aetiology

- Degenerative mitral valve disease. Commonest cause in West.

Represents spectrum from fibroelastic deficiency (small valves, single segment prolapse), to Barlow's disease (large valves, multisegment prolapse). Also called floppy valve, prolapse, myxomatous disease.

- Rheumatic heart disease.
- Infective endocarditis
- Connective tissue disorders, e.g. Marfan and Ehlers–Danlos syndromes.
- Ischemic heart disease.
- Congenital cleft valve leaflet (associated with primum ASD).
- Endomyocardial fibrosis (common in sub-Saharan Africa).
- Iatrogenic (balloon valvuloplasty of stenotic valve).
- Prosthesis failure (paraprosthetic leak, leaflet perforation).

Tricupid valve disease

TS is rare—usually rheumatoid. TR may be caused by:

- *Functional* secondary to mitral valve disease (commonest).
- *Rheumatic* heart disease.
- *Infective endocarditis.*
- Ebstein's anomaly.
- *Carcinoid* syndrome (usually associated with pulmonary regurgitation).
- Endomyocardial fi brosis.
- Prolapsing cusp.

PULMONARY VALVE DISEASE

Acquired pulmonary valve disease is unusual. Severe pulmonary hypertension may cause dilatation of the pulmonary valve ring causing pulmonary regurgitation.

TYPES OF VALVE PROSTHESIS

Key facts

• Valve prostheses are either *mechanical* or *bioprosthetic* ('tissue').

• Tissue valves are mounted on a metal frame (*stented*), or supported by pig aorta and cloth (*stentless)*. Stented most commonly used.

• Stented valves are either *porcine* aortic valves or *bovine* pericardium.

• *Homografts* are human cadaveric aortic roots, complete with aortic valves *in situ* .

• A *pulmonary autograft* is the patient's own excised pulmonary valve used in the Ross procedure