CONGENITAL HEART DISEASE

Congenital heart disease usually manifests in childhood but may pass unrecognised and not present until adult life. Patients may remain well for many years and subsequently re-present in later life with related problems such as arrhythmia,valv. HD., or ventricular dysfunction .

The fetal circulation allows oxygenated blood from the placenta, through umdelicar artery ,to IVC, to RA., to pass directly to the left side of the heart through the foramen ovale without having to flow predominantly through RV.,then pulm. A.,to descending aorta , not to the lungs, through Ductus arteriosus, since pulmonary vasculature has a high resistance .

At birth, the lungs expand with air and pulmonary vascular resistance falls so that blood now flows to the lungs and back to the left atrium. left atrial pressure rises above right atrial pressure and the flap valve of the foramen ovale closes.

Atrial septal defects occur at the site of the foramen ovale. A patent ductus arteriosus may remain if it fails to close after birth. Failure of the aorta to develop at the point of the aortic isthmus and where the ductus arteriosus attaches can lead to narrowing or coarctation of the aorta.

Down's syndrome may cause septal defects. Cerebrovascular accidents and cerebral abscesses are complications of severe cyanotic congenital disease.

Early diagnosis is important because many types of congenital heart disease are amenable to surgical treatment, but this opportunity may be lost if secondary changes such as pulmonary vascular damage occur, within years(Eisenmenger syndrome).

Presentations: Neonatal central cyanosis , HF (peripheral cyanosis is harmless, and not related). Childhood central cyanosis / HF. /Failure to thrive –growth retardation /tachyarrhythmias.Adulthood HF./Eisenmengers syndrome / secondaryv HT., in coarctation / related murmurs/ related post operative corrective open heart surgeries(tachyarrhythmias ,HF. Infections).

Central cyanosis and digital clubbing

Central cyanosis of cardiac origin occurs when desaturated blood enters the systemic circulation without passing through the lungs (i.e. a right-to-left shunt). In the neonate, the most common cause is transposition of the great arteries, in which the aorta arises from the right ventricle and the pulmonary artery from the left. In older children, cyanosis is usually the consequence of a ventricular septal defect combined with severe pulmonary stenosis (tetralogy of Fallot) or with pulmonary vascular disease (Eisenmenger's syndrome). Prolonged cyanosis is associated with finger and toe clubbing (with one exception: reverced central cyanosis and differential clubbing ; PHT. In a large PDA.).

Growth retardation and learning difficulties

These may be a feature with large left-to-right shunts at ventricular or great arterial level, if they form part of a genetic syndrome as Dowen syndrome,or rarely as sequelae of cerebral embolism in reversed shunting.

Syncope

In the presence of increased pulmonary vascular resistance or severe left or right ventricular outflow obstruction, exercise may provoke syncope as systemic vascular resistance falls on exercise but pulmonary vascular resistance may rise, worsening right-to-left shunting and cerebral oxygenation.

Pulmonary hypertension and Eisenmenger's syndrome

Persistently raised pulmonary flow (e.g. with left-to-right shunt) leads to increased pulmonary resistance followed by pulmonary hypertension., once established, the increased pulmonary resistance is irreversible. Central cyanosis appears and digital clubbing develops. The chest X-ray shows enlarged central pulmonary arteries and peripheral 'pruning' of the pulmonary

vessels. cardiac failure may eventually ensue, dyspnoea being the first symptom. A continuous 'machinery' murmur is heard with late systolic accentuation, maximal in the second left intercostal space below the clavicle .It is frequently accompanied by a thrill.The ECG shows right ventricular hypertrophy. If severe pulmonary hypertension develops, a left-to-right shunt may reverse, resulting in right-to-left shunting and marked cyanosis (Eisenmenger's syndrome). This is more common with large ventricular septal defects or persistent ductus arteriosus than with atrial septal defects(which might occur lately in elderlies). Patients with Eisenmenger's syndrome are at particular risk from abrupt changes in afterload that exacerbate right-to-left shunting, e.g. vasodilatation, anaesthesia, pregnancy.

Pregnancy

During pregnancy, there is a 50% increase cardiac output. Abnormalities causing severe outflow tract obstruction, such as aortic stenosis, are not well tolerated and are associated with significant maternal morbidity and mortality. Most patients with surgically corrected congenital heart disease, and many with palliated or untreated disease, will tolerate pregnancy well. Pregnancy is particularly hazardous in the presence of conditions associated with cyanosis or severe pulmonary hypertension. For example, maternal mortality in patients with Eisenmenger's syndrome is more than 50% .

Patent ductus arteriosus -PDA.

With small shunts there may be no symptoms for years. If large defect: HF may eventually ensue, dyspnoea being the first symptom. A continuous 'machinery' murmur is hear, maximal in the second left intercostal space below the clavicle, with a local thrill.If coplicated by PHT. ,shunt through the defect may then reverse, causing central cyanosis (Eisenmenger's syndrome), which may be more apparent in the feet and toes than in the upper part of the body. The murmur becomes quieter, may be confined to systole or may disappear. The ECG shows evidence of right ventricular hypertrophy.
It is now usual practice to close a patent ductus at cardiac catheterisation with an implantable occlusive device. Closure should be undertaken in infancy if the shunt is significant and pulmonary resistance not elevated.

COARCTATION OF THE AORTA

Narrowing of the aorta most commonly occurs in the region where the ductus arteriosus joins the aorta, i.e. at the isthmus just below the origin of the left subclavian artery .
It may be associated with bicuspid aortic valve.
Headaches may occur from hypertension ;weakness or cramps in the leg; The blood pressure is raised in the upper body but normal or low in the legs; The femoral pulses are weak, and delayed in comparison with the radial pulse ; A systolic murmur is usually heard posteriorly, over the coarctation; There may also be an ejection click and systolic murmur in the aortic area due to a bicuspid aortic valve.
Chest X-ray at a later age may show changes in the contour of the aorta (indentation of the descending aorta, '3 sign') ; notching of the under-surfaces of the ribs from collaterals ;MRI is ideal for demonstrating the lesion ;ECG left ventricular hypertrophy.
Surgical correction is advisable in all but the mildest cases. If this is done sufficiently early in childhood, persistent hypertension can be avoided. Patients repaired in late childhood or adult life often remain hypertensive or develop recurrent hypertension later in life. Recurrence of stenosis may occur as the child grows, and this may be managed by balloon dilatation, which can also be used as the primary treatment in some cases.

Atrial septal defect –ASD.

ATRIAL SEPTAL DEFECT

Most are 'ostium secundum' defects, involving the fossa ovalis which in utero was the foramen ovale). 'Ostium primum' defects result from a defect in the atrioventricular septum and are associated with a 'cleft mitral valve' (split anterior leaflet).

There is gradual enlargement of the right side . Pulmonary hypertension and shunt reversal are less common and tend to occur later in life than with other types of left-to-right shunt.

Most children are free of symptoms for many years and the condition is often detected at routine clinical examination or following a chest X-ray. Dyspnoea, recurrent chest infections, and arrhythmias, especially atrial fibrillation, as late presentations. The characteristic physical signs are the result of the volume overload of the right ventricle:

- wide fixed splitting of the second heart sound: wide because of delay in right ventricular ejection (increased stroke volume and right bundle branch block) and fixed because the septal defect equalises left and right atrial pressures throughout the respiratory cycle
- a systolic flow murmur over the pulmonary valve, mimiking valv.PS, but with fixed splitting of loud S2 .

The chest X-ray typically shows enlargement of the heart and the pulmonary artery as well as pulmonary plethora; ECG :incomplete right bundle branch block ;Echocardiography :demonstrate the defect and shows RV dilatation, RV hypertrophy and pulmonary artery dilatation; The precise size and location of the defect can be shown by transoesophageal echocardiography.

Closure can be accomplished at cardiac catheterisation using implantable closure devices,rather than surgery if there are adequate rims or not primumum type(surgery). Severe pulmonary hypertension and shunt reversal are both contraindications to attempted closure.

VENTRICULAR SEPTAL DEFECT

Embryologically, the interventricular septum has a membranous and a muscular portion, and the latter is further divided into inflow(near TV.), trabecular (muscular), and outflow portions(near PV.) . Most congenital defects are 'perimembranous', i.e. at the junction of the membranous and muscular portions.

The defect may be isolated or part of complex congenital heart disease. Acquired ventricular septal defect may result from rupture as a complication of acute myocardial infarction.

Flow from the high-pressure left ventricle to the low-pressure right ventricle during systole produces a pansystolic murmur usually heard best at the left sternal edge but radiating all over the precordium. A small defect often produces a loud murmur (maladie de Roger) in the absence of other haemodynamic disturbance.

Conversely, a large defect may produce a softer murmur, particularly if pressure in the right ventricle is elevated producing prominent parasternal pulsation, tachypnoea and indrawing of the lower ribs on inspiration. The chest X-ray shows pulmonary plethora and the ECG shows bilateral ventricular hypertrophy.

May present as cardiac failure in infants; as a murmur with only minor haemodynamic disturbance in older children or adults; or rarely as Eisenmenger's syndrome. In a proportion of

infants, the murmur gets quieter or disappears due to spontaneous closure of the defect, esp. trabecular muscular type.
Small ventricular septal defects require no specific treatment apart from endocarditis prophylaxis.
Cardiac failure caused by a ventricular septal defect in infancy is initially treated medically with digoxin and diuretics. Persisting failure is an indication for surgical repair of the defect. Percutaneous closure devices are under development.
Many patients with Eisenmenger's syndrome die in the second or third decade of life, but a few survive to the fifth decade without transplantation.

TETRALOGY OF FALLOT
The tetralogy comprises (1) pulmonary stenosis(subvalvular -infundibular), (2) overriding of the ventricular septal defect by the aorta, (3) a ventricular septal defect (large and similar in aperture to the aortic orifice).and (4) right ventricular hypertrophy .Combination results in elevated right ventricular pressure and right-to-left shunting of cyanotic blood across the ventricular septal defect, leading to dilated AA. .
Children are usually cyanosed but this may not be present in the neonate because it is only when right ventricular pressure rises to equal or exceed left ventricular pressure that a large right-to-left shunt develops.
Subvalvular component of the right ventricle outflow obstruction is dynamic, and may increase suddenly under adrenergic stimulation. The affected child suddenly becomes increasingly cyanosed, often after feeding or a crying attack, and may become apnoeic and unconscious. These attacks are called 'Fallot's spells. In older children, Fallot's spells are uncommon but cyanosis becomes increasingly apparent, with stunting of growth, digital clubbing and polycythaemia. Some children characteristically obtain relief by squatting after exertion, which increases the afterload of the left heart and reduces the right-to-left shunting.
The most characteristic feature is the combination of cyanosis with a loud ejection systolic murmur in the pulmonary area (as for pulmonary stenosis). Cyanosis may be absent in the newborn or in patients with only mild right ventricular outflow obstruction ('acyanotic tetralogy of Fallot').
ECG: right ventricular hypertrophy; chest X-ray :small pulmonary artery ,a 'boot-shaped' heart:Echocardiography : diagnostic : the aorta is not continuous with the anterior ventricular septum.
Primary surgical correction may be undertaken prior to age 5, unless the pulmonary arteries are too hypoplastic, when a palliative shunt may be performed (e.g. the Blalock-Taussig shunt, an anastomosis between the pulmonary artery and subclavian artery). This improves pulmonary blood flow and pulmonary artery development, and may facilitate definitive correction at a later stage. Follow-up is needed to identify residual shunting, recurrent pulmonary stenosis, significant apr., and rhythm disorders(VT.).
Other causes of cyanotic congenital H. dis. : tricuspid atresia (since delivery, with atretic RV., and shunting)/ transposition of great vessels(deep central cyanosis , since delivery, with shunting, those who have had a 'Mustard' repair, where blood is re-directed at atrial level leaving the right ventricle connected to the aorta, may develop right ventricular failure in adult life. /PA. atresia(deep cyanosis since delivery /Ebestiens anomaly of TV.(central cyanosis, lately, wit syncopal attacks duecto tachyarrhythmias)

Those who have had surgery involving the atria may develop atrial arrhythmias, and those who have ventricular scars may develop ventricular arrhythmi