CONGENITAL HEART DISEASE

EPIDEMIOLOGY

- Congenital heart disease occurs in approximately 0.8% of live births.
- The incidence is higher in stillborns (3-4%), spontaneous abortus (10-25%), and premature infants (about 2% excluding PDA).
- Most cases are multifactorial but some are associated with chromosomal disorders, single gene defects, teratogens or maternal metabolic disease.

Table 418-1 RELATIVE FREQUENCY OF MAJOR CONGENITAL HEART LESIONS*

LESION	% OF ALL LESIONS
Ventricular septal defect	35-30
Atrial septal defect (secundum)	6-8
Patent ductus arteriosus	6-8
Coarctation of aorta	5-7
Tetralogy of Fallot	5-7
Pulmonary valve stenosis	5-7
Aortic valve stenosis	4-7
d-Transposition of great arteries	3-5
Hypoplastic left ventricle	1-3
Hypoplastic right ventricle	1-3
Truncus arteriosus	1-2
Total anomalous pulmonary venous return	1-2
Tricuspid atresia	1-2
Single ventricle	1-2
Double-outlet right ventricle	1-2
Others	5-10

*Excluding patent ductus arteriosus in preterm neonates, bicuspid aortic valve, physiologic peripheral pulmonic stenosis, and mitral valve prolapse.

- Congenital cardiac defects can be divided into 2 major groups based on the presence or absence of cyanosis, which can be determined by physical examination aided by pulse oximetry.

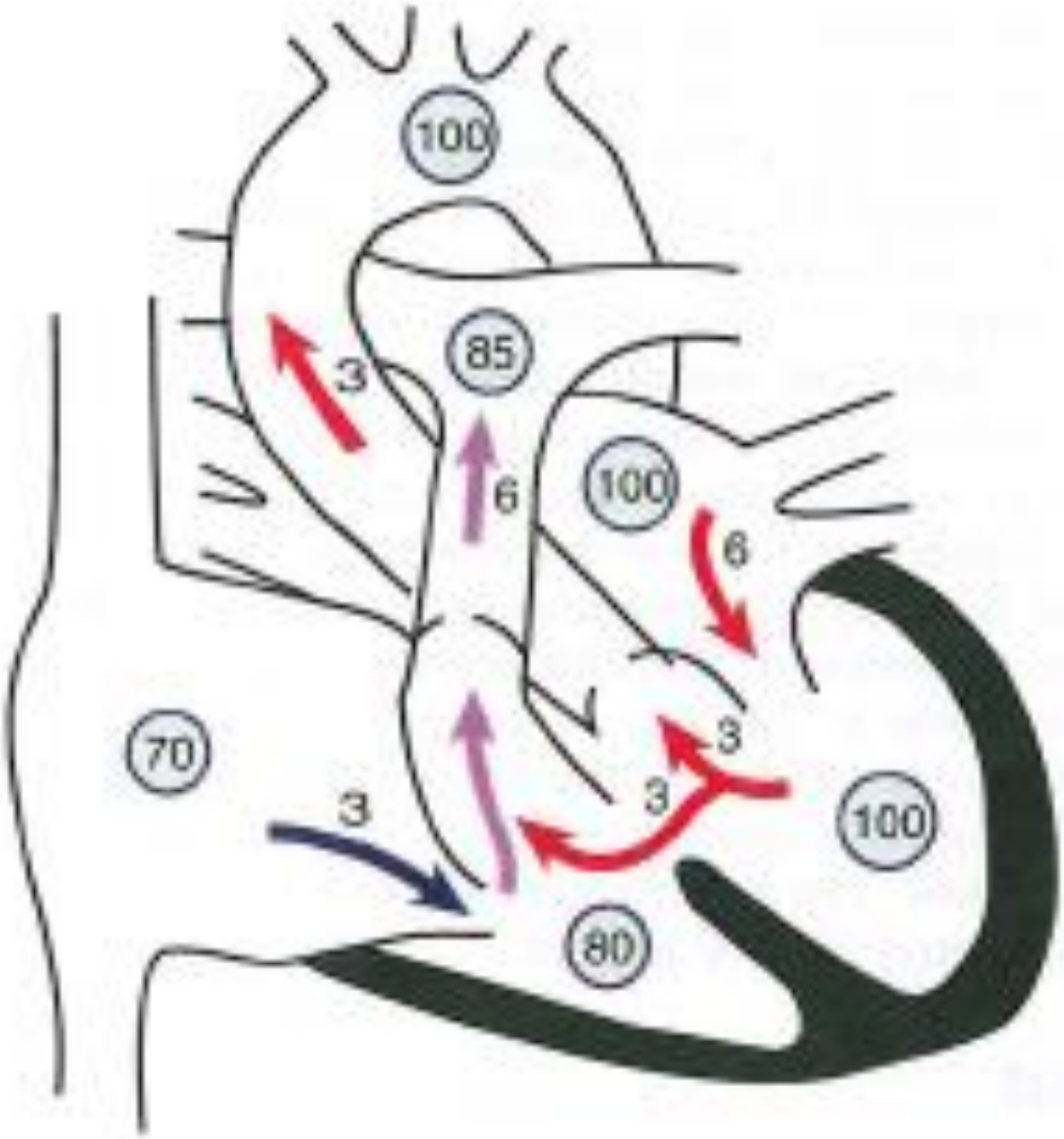
ACYANOTIC CONGENITAL HEART LESIONS

- Acyanotic congenital heart lesions can be classified according to the predominant physiologic load that they place on the heart.
- The most common lesions are those that produce a volume load, and the most common of these are left-to-right shunt lesions.

- The second major class of lesions causes an increase in pressure load, most commonly secondary to ventricular outflow obstruction (pulmonic or aortic valve stenosis) or narrowing of one of the great vessels (coarctation of the aorta).

Ventricular Septal Defect

- The most common congenital heart defect (25%).
- Muscular, inlet, supracristal and membranous defects
- Perimembranous VSDs are the most common
- Shunt depends on the size of the defect and the pulmonary vascular resistance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Clinical manifestations

- Small VSDs, often asymptomatic but a loud murmur
- Moderate to large VSDs, pulmonary overcirculation and heart failure, presenting as fatigue, diaphoresis with feeding and poor growth, a pansystolic murmur at the lower left sternal border
- Large shunts, a mid-diastolic murmur at the apex.

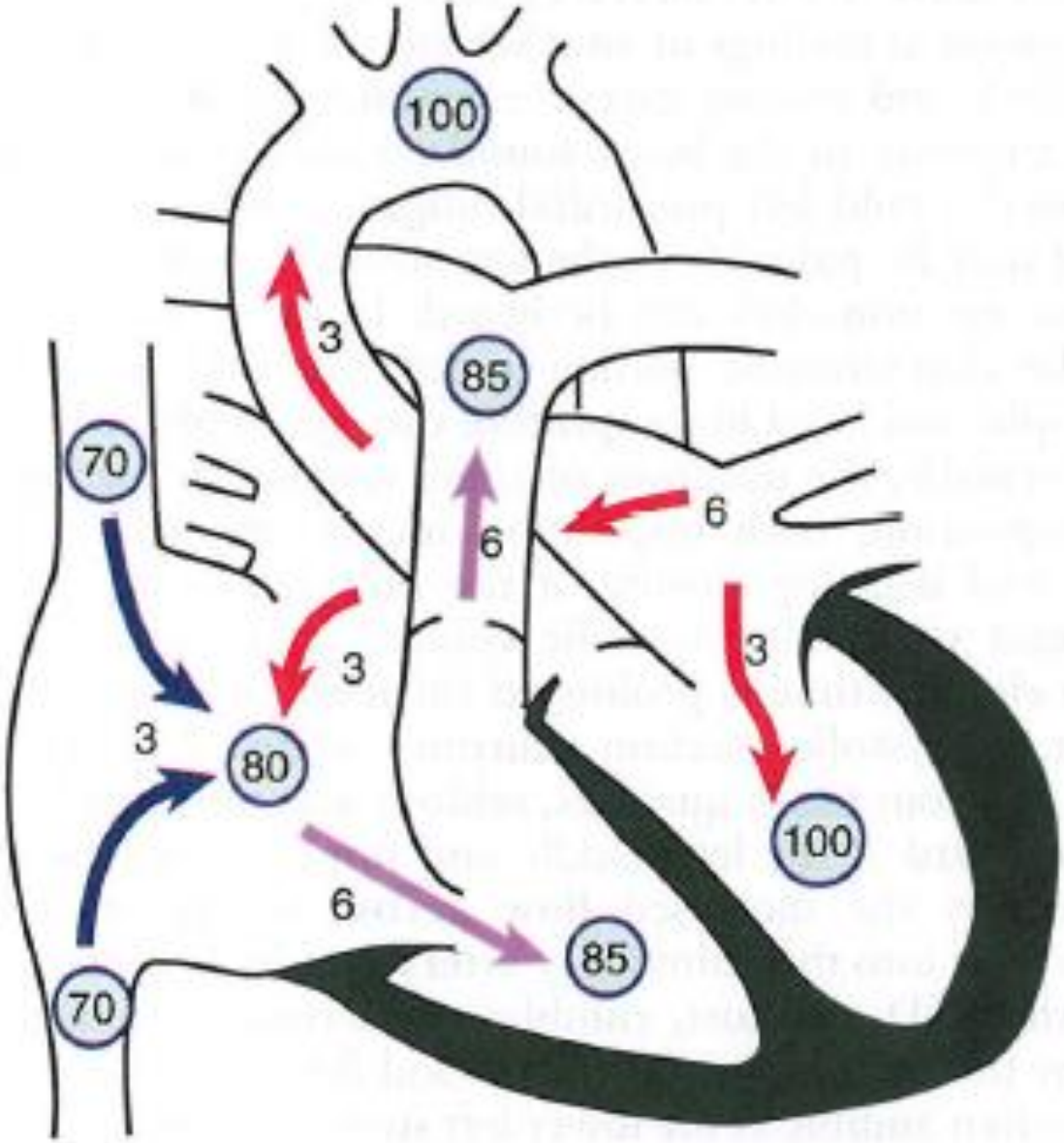
- ECG is normal with small VSDs but LA enlargement and LV hypertrophy with large VSDs.
- Chest X ray may reveal cardiomegaly, increase in pulmonary artery silhouette and pulmonary blood flow, and finally RV enlargement.

Treatment

- One thirds close spontaneously.
- Small VSDs usually close spontaneously and if they do not, surgical closure may not be required.
- Moderate to large VSDs usually need medical treatment including diuretics, digoxin and afterload reduction.
- Poor growth or pulmonary hypertension requires closure (surgical or with device).

Atrial Septal Defect

- Failure of septal growth or excessive resorption of tissue
- 10% of all CHDs
- Secondum, primum and sinus venosus defects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Clinical manifestations

- Shunt depends on size of the defect and the compliance of the both ventricles
- A prominent RV impulse at the lower LSB, a soft systolic ejection murmur in the region of RVOT, a fixed split S2
- A large shunt can result in a mid-diastolic murmur at the LSB

- ECG may show RAD and RVH
- Chest x-ray may show RAE and prominent PA

Treatment

- If there is a significant shunt, closure at 3 years of age (with device or surgically)

Patent Ductus Arteriosus

- Failure of the normal closure after birth
- 5-10% of CHDs

Clinical manifestations

- Shunt depends on the size of PDA and the pulmonary vascular resistance
- Moderate to large shunts produce heart failure
- A wide pulse pressure, a continuous murmur at the left infraclavicular area
- Larger shunts can result in a mid-diastolic murmur at the apex and a hyperdynamic precordium

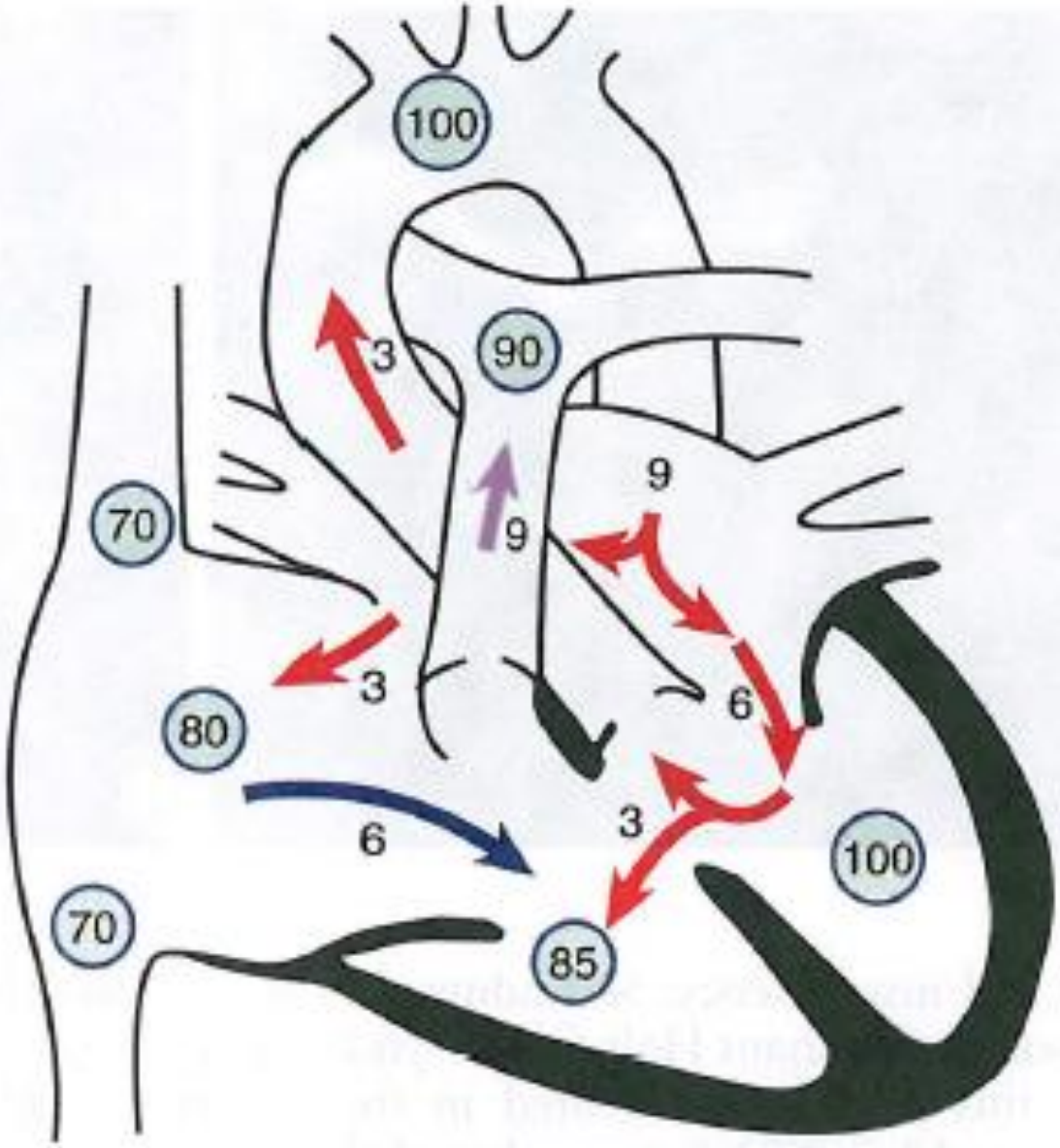
- ECG may show LVH, and RVH(PH)
- Chest x-ray may show full pulmonary artery silhouette and increased pulmonary vascularity

Treatment

- Spontaneous closure in term infants is uncommon after a few weeks
- For hemodynamically insignificant PDAs, controversial (closure with device or surgically)
- Moderate to large PDAs, medical management and then closure

Endocardial Cushion Defect

- Abnormal development of endocardial cushion tissue
- The complete defect results in a primum ASD, an inlet VSD and clefts in the anterior leaflet of the mitral and septal leaflet of the tricuspid valves
- Left to right Shunting at both levels, AV valve insufficiency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Clinical manifestations

- Symptoms of heart failure at the first 6 to 8 weeks of life
- Pulmonary hypertension, poor growth, murmurs, single S2
- Down syndrome may accompany CAVSD

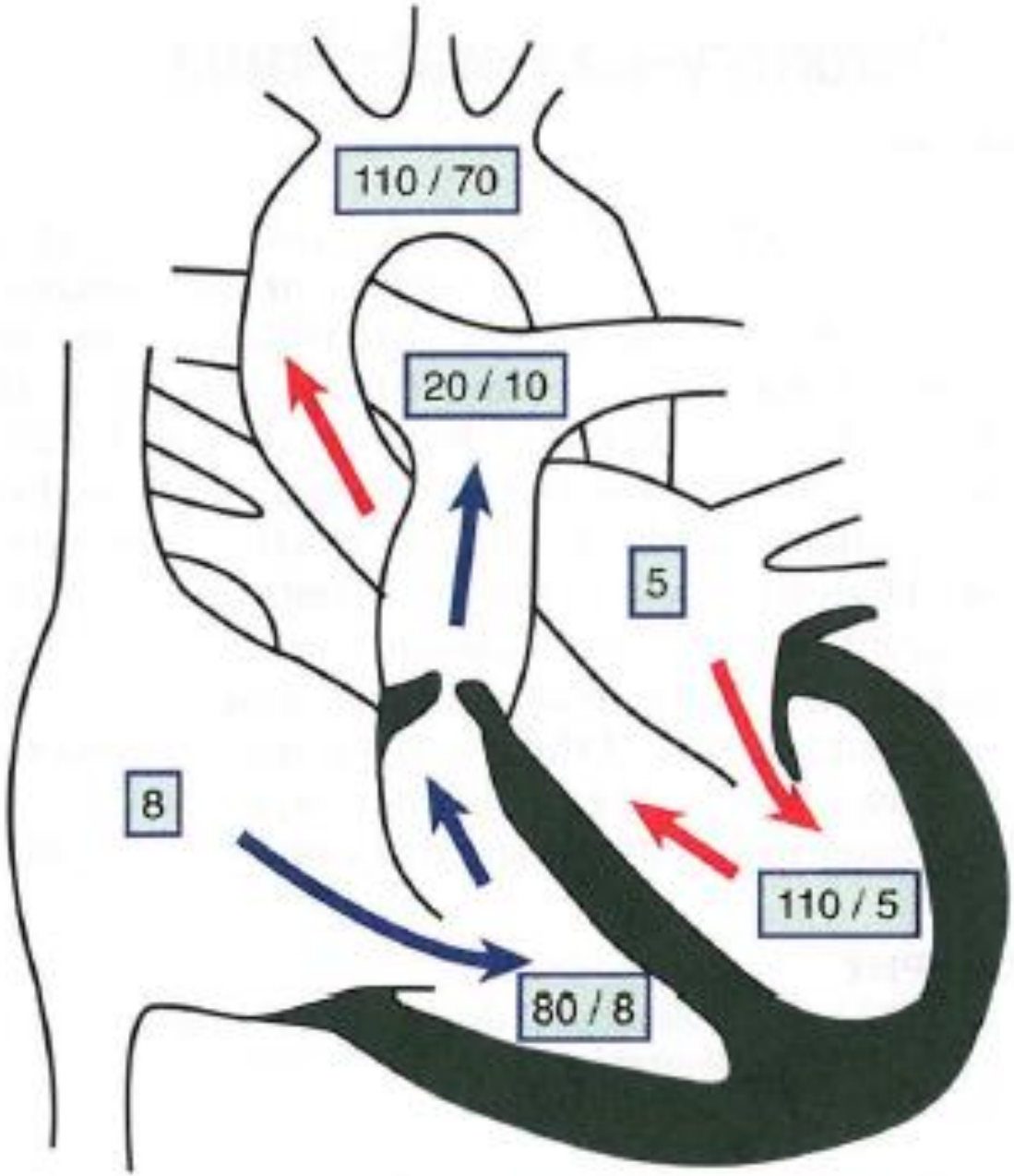
- ECG reveal LAD and combined ventricular hypertrophy, may show combined atrial enlargement
- Chest x-ray reveals cardiomegaly and increased vascularity
- Echocardiography shows the diagnosis

Treatment

- Medical treatment (diuretics, digoxin and afterload reducers)
- Surgical repair ultimately

Pulmonary Stenosis

- Failure of the development of the three leaflets, insufficient resorption of infundibular septum or insufficient canalization of the peripheral pulmonary arteries in early gestation
- 10% of CHDs
- Valvular, subvalvular or supravalvular


110 / 70
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8
110 / 5
80 / 8

Clinical manifestations

- Mild stenosis is asymptomatic
- Moderate to severe stenosis produces exertional dyspnea, easy fatigability
- In newborns, cyanosis
- Systolic ejection murmur at the second left intecostal space, thrill, widely split S2, LSB impulse, click varying with respiration
- Increase in duration and frequency of murmurs with worsening of stenosis

- ECG shows RAD and RVH with moderate to severe stenosis
- Chest x-ray usually shows normal size heart, may show post-stenotic dilatation of the main pulmonary artery as prominent PA segment
- Echocardiography reveals site of the stenosis, degree of hypertrophy, valve morphology and estimates pressure gradient

Treatment

- Valvular pulmonary stenosis does not progress
- Balloon valvuloplasty for more significant or symptomatic stenosis
- Surgical repair for unsuccessful valvuloplasty or subvalvular stenosis

Aortic Stenosis

- 5% of all CHDs
- Failure of development of the three leaflets or resorption of tissue around the valve
- Valvular, subvalvular and supravalvular

Clinical manifestations

- Moderate to severe stenosis produce easy fatigability, exertional chest pain and syncope
- Infants may present with heart failure (critical AS)
- A systolic ejection murmur at the right second intercostal space, a systolic ejection click, a thrill at upper RSB or suprasternal notch
- Increase in duration and frequency of murmurs with worsening of stenosis

- ECG shows LVH with moderate to severe stenosis
- Chest x-ray shows LVE with moderate to severe stenosis, may reveal post-stenotic dilatation of the ascending aorta or aortic knob
- Echocardiography reveals site of the stenosis, valve morphology, LV hypertrophy and estimates pressure gradient

Treatment

- The degree of stenosis frequently progresses with growth and age
- Balloon valvuloplasty for more significant or symptomatic stenosis
- Surgical repair for unsuccessful valvuloplasty or significant valve insufficiency

Coarctation of the Aorta

- 10% of all CHDs
- Juxtaductal and infantile types

Clinical manifestations

- Infants frequently have hypoplastic arches, associated lesions and may be dependent to PDA to provide descending aorta flow
- Poor feeding, respiratory distress and shock, radiofemoral delay, lower extremity blood pressure, S3
- Older children are asymptomatic, history of leg discomfort, headache, epistaxis, decreased or absent lower extremity pulses, upper extremity hypertension, murmur in the left interscapular area

- ECG and CXR show RVH, marked cardiomegaly and pulmonary edema in infants, LVH and mild enlarged heart, rib notching in older children
- Echocardiography shows the site and the degree of coarctation, LVH and associated lesions

Treatment

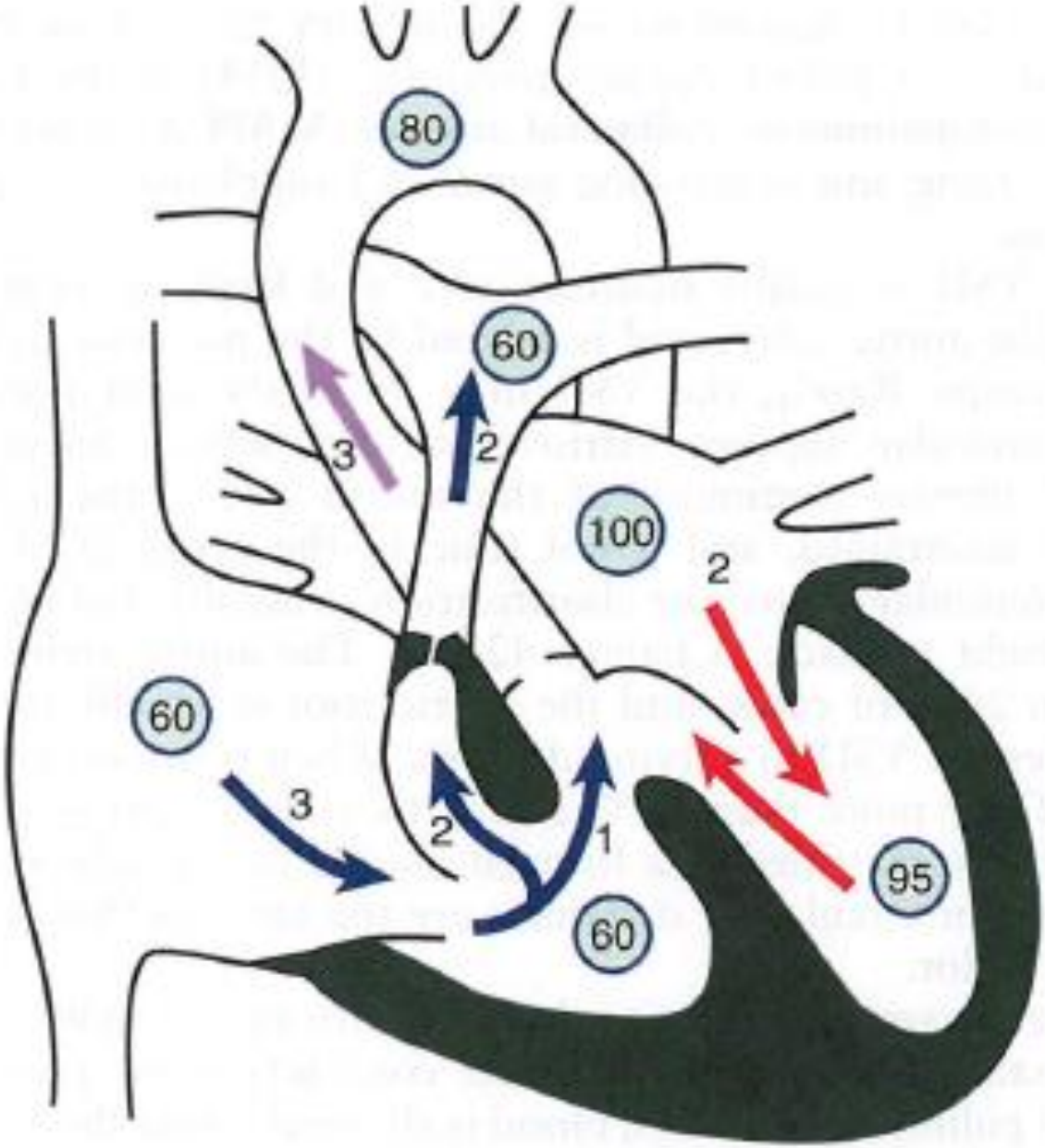
- Infusion of PGE1, inotropes, diuretics and then balloon angioplasty or surgical repair for infants
- Balloon angioplasty or stenting and also surgical repair for older children

CYANOTIC CONGENITAL HEART LESIONS

- Some of the systemic venous return returns to the body without going through the lungs
- Cyanosis, 5gr/dl of reduced hemoglobin

Tetralogy of Fallot

- The most common cyanotic CHD, 10% of all CHDs
- Includes VSD, PS, overriding aorta and RVH
- Abnormal septation of the truncus arteriosus in early gestation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Clinical manifestations

- The degree of cyanosis depends on the amount of pulmonary stenosis
- A murmur, a single S2, RV impulse at the LSB
- Tet spells
- Cerebral thromboembolism and abscess

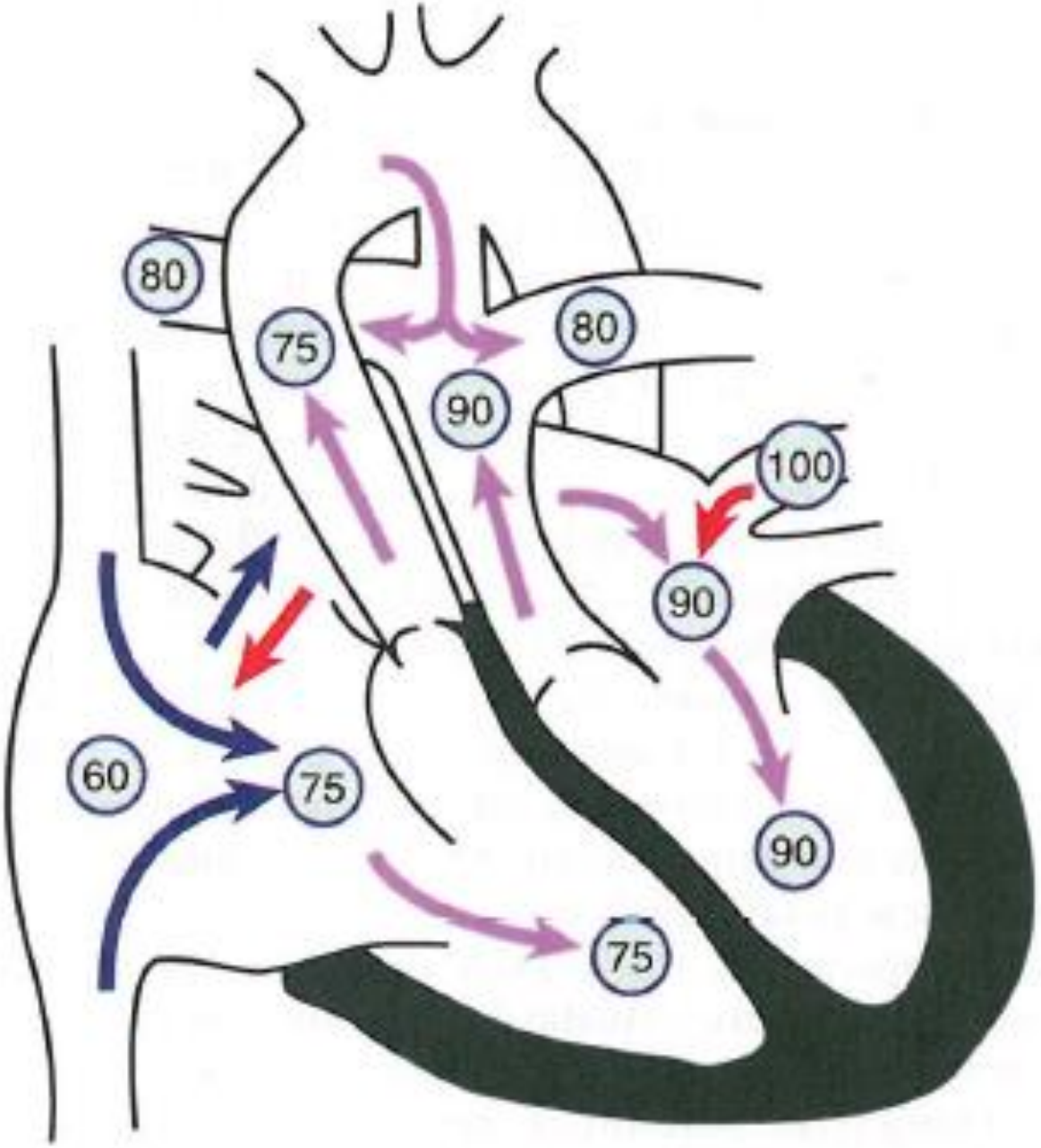
- ECG shows RAD and RVH
- Chest x-ray shows boot-shaped heart
- Echocardiography shows the anatomic features

Treatment

- Tet spells: O2, chest-knee position, MS, α adrenergic agonist
- Surgical repair
- Palliative shunt surgery
- SBE prophylaxis

Transposition of the Great Arteries

- 5% of all CHDs
- The most cyanotic lesion presenting in newborn period
- Abnormality of septation of the truncus arteriosus
- Desaturated blood returns to the right side and saturated blood to the left side of the heart, without mixing (ASD, PFO, VSD, PDA) death occurs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Clinical manifestations

- Cyanosis, quiet tachypnea, single S2
- Murmur, less cyanosis, signs of heart failure if VSD presents

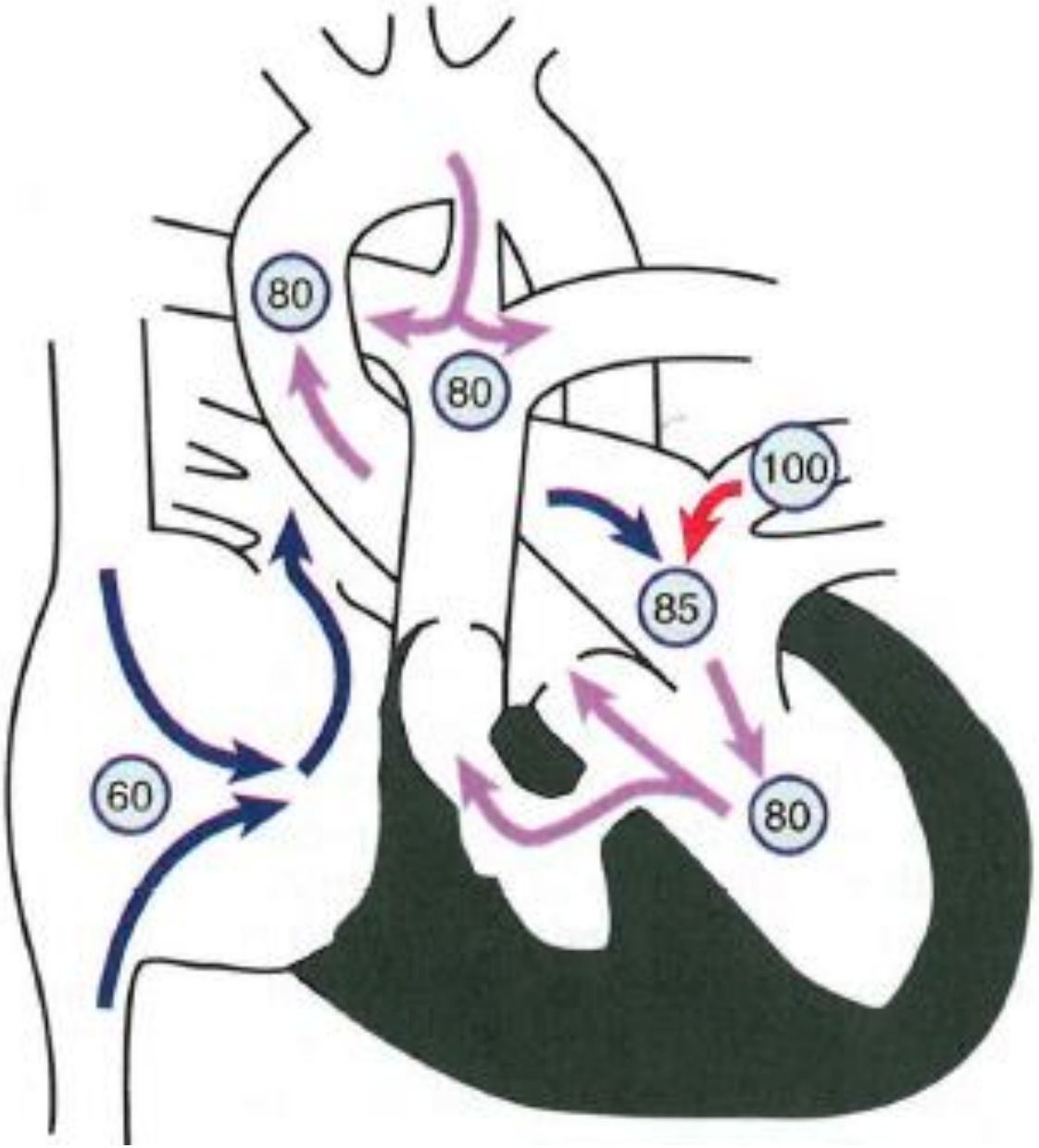
- ECG shows RAD, RVH
- Chest x-ray shows egg on a string, increased pulmonary vascularity
- Echocardiography reveals the diagnosis and associated lesions

Treatment

- PGE1
- Balloon atrial septostomy (persistent significant hypoxia)
- Surgical repair

Tricuspid Atresia

- 2% of all CHDs
- Failure in normal development of the valve from endocardial cushions and septal tissue
- Hypoplastic RV
- A PDA or VSD is necessary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Clinical manifestations

- Severe cyanosis, single S2
- Murmur of the VSD, diastolic murmur across the mitral valve

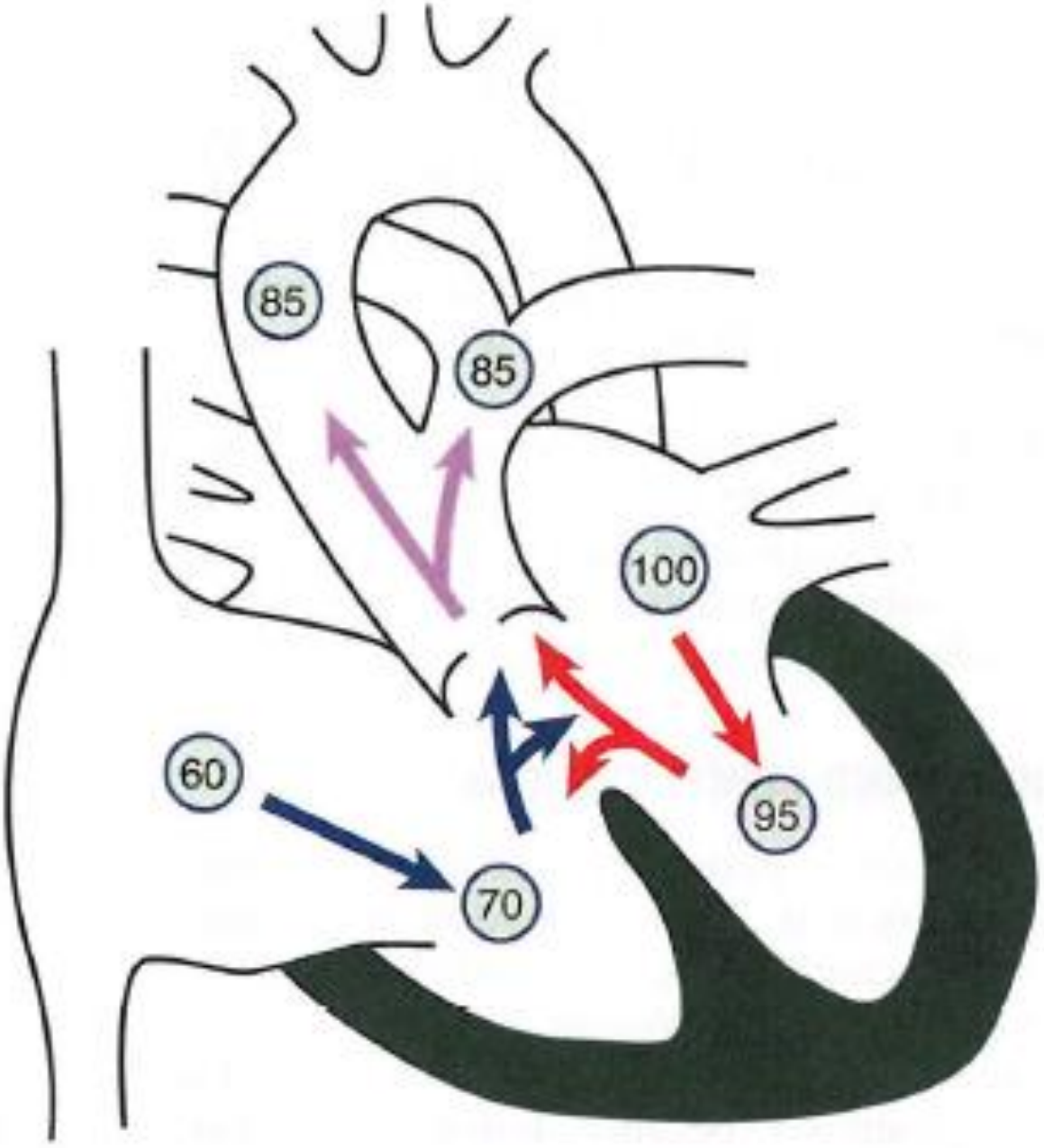
- ECG shows LVH, superior QRS axis
- Chest x-ray shows normal or mildly enlarged heart , decreased pulmonary blood flow
- Echocardiography shows the anatomy, associated lesions, source of pulmonary blood supply

Treatment

- PGE1, if there is no VSD
- Surgical repair

Truncus Arteriosus

- Less than 1% of CHDs
- Failure of the septation of the truncus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Clinical manifestations

- Cyanosis, heat failure, bounding peripheral pulses, single S2, systolic ejection click, SMM at the LSB

- ECG shows combined ventricular hypertrophy
- Chest x-ray shows increased pulmonary flow, displaced pulmonary arteries
- Echocardiography defines the anatomy

Treatment

- Medical management
- Surgical repair

Total Anomalous Pulmonary Venous Return

- 1% of CHDs
- Disruption of the development of normal pulmonary venous drainage
- All of the pulmonary veins return abnormally via the right side of the heart
- Supracardiac, cardiac, infracardiac and mixed drainage
- An ASD is required for survival

Clinical manifestations

- Without obstruction, asymptomatic, RV impulse, widely split S2, SMM at the upper LSB, mid-diastolic murmur at the lower LSB
- With obstruction, cyanosis, tachypnea and dyspnea, right sided heart failure, no murmur

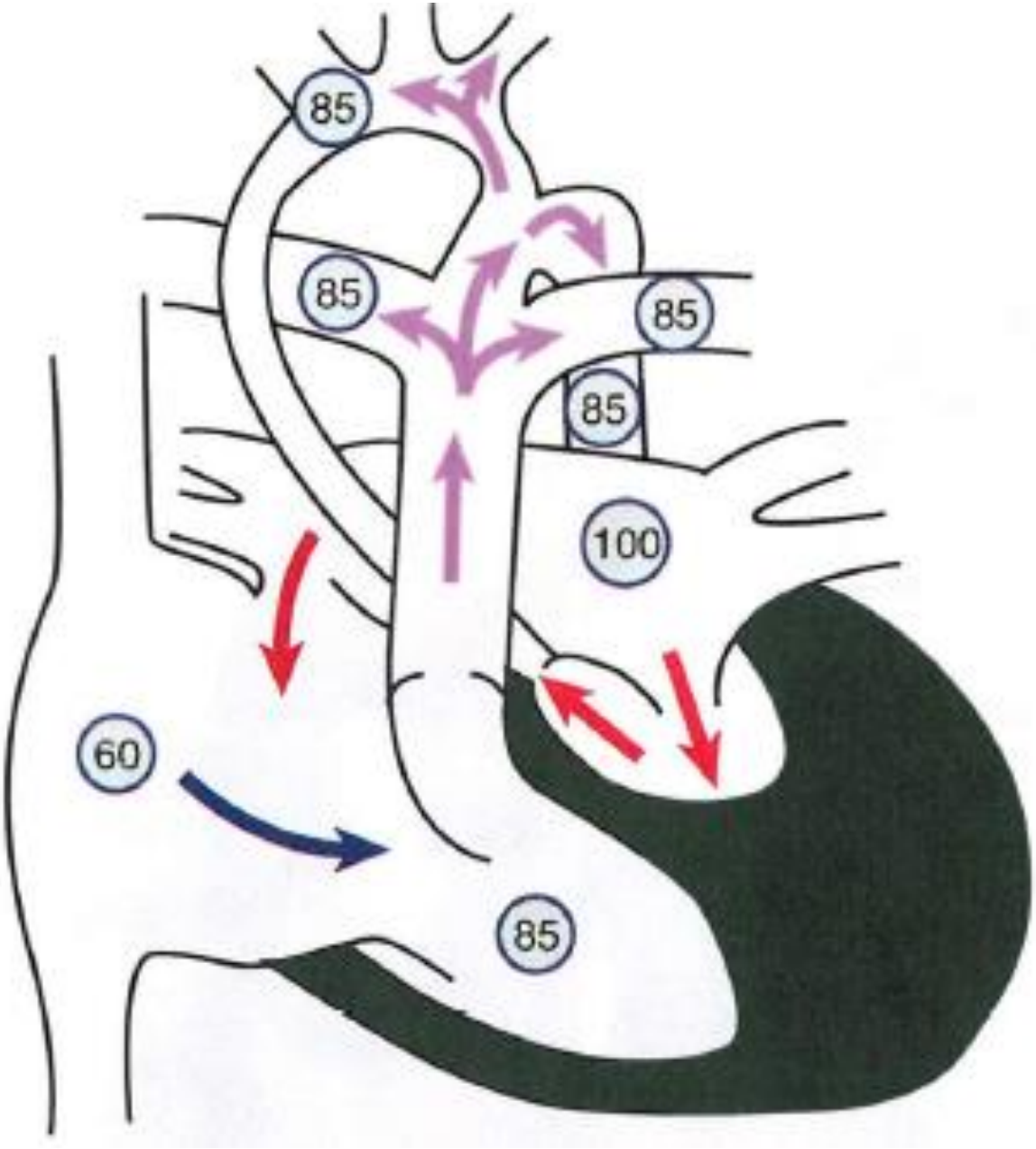
- ECG shows RV overload without obstruction and RAD, RVH with obstruction
- Chest x-ray shows cardiomegaly and increased pulmonary blood flow without obstruction and normal or mildly enlarged heart, pulmonary edema with obstruction
- Echocardiography shows right side volume overload, right to left atrial shunt, common pulmonary vein drainage and degree of obstruction

Treatment

- Surgical repair

Hypoplastic Left Heart Syndrome

- 1% of CHDs
- The most common cause of death in the first month of life
- Failure of the development of mitral or aortic valve or aortic arch
- A small LV unable to support normal circulation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Clinical manifestations

- As the ductus constricts, illness, heart failure, diffusely weak pulses, single S2, usually no murmur, cyanosis, grayish color

- ECG shows RVH, decreased LV forces
- Chest x-ray shows cardiomegaly, pulmonary edema
- Echocardiography shows small left heart, stenotic MV and AO valve, hypoplastic arch, left to right atrial shunt

Treatment

- PGE1, correction of acidosis, blood pressure and ventilatory support
- Surgical repair

TABLE 144-1 Categories of Presenting Symptoms and Signs in the Neonate

Symptom/Sign	Physiologic Category	Anatomic Cause	Lesion
Cyanosis with respiratory distress	Increased pulmonary blood flow	Transposition	*d*-Transposition with or without associated lesions
Cyanosis without respiratory distress	Decreased pulmonary blood flow	Right heart obstruction	Tricuspid atresia Ebstein anomaly Pulmonary atresia Pulmonary stenosis Tetralogy of Fallot
Hypoperfusion	Poor cardiac output	Left heart obstruction	Total anomalous pulmonary venous return with obstruction Aortic stenosis Hypoplastic left heart syndrome
	Poor cardiac function	Normal anatomy	Cardiomyopathy Myocarditis
Respiratory distress with desaturation (not visible cyanosis)	Bidirectional shunting	Complete mixing	Truncus arteriosus AV canal Complex single ventricle (including heterotaxias) without pulmonary stenosis
Respiratory distress with normal saturation	Left-to-right shunting	Simple intracardiac shunt	ASD VSD PDA Aortopulmonary window AVM

Complications of CHDs

TABLE 144-2 Extracardiac Complications of Cyanotic Congenital Heart Disease

Problem	Etiology	Therapy
Polycythemia	Persistent hypoxia	Phlebotomy
Relative anemia	Nutritional deficiency	Iron replacement
CNS abscess	Right-to-left shunting	Antibiotics, drainage
CNS thromboembolic stroke	Right-to-left shunting or polycythemia	Phlebotomy
Gingival disease	Polycythemia, gingivitis, bleeding	Dental hygiene
Gout	Polycythemia, diuretic agents	Allopurinol
Arthritis, clubbing	Hypoxic arthropathy	None
Pregnancy	Poor placental perfusion, poor ability to increase cardiac output	Bed rest
Infectious disease	Associated asplenia, DiGeorge syndrome	Antibiotics
	Fatal RSV pneumonia with pulmonary hypertension	Ribavirin, RSV immune globulin
Growth	Failure to thrive, increased oxygen consumption, decreased nutrient intake	Treat heart failure; correct defect early
Psychosocial adjustment	Limited activity, peer pressure; chronic disease, multiple hospitalizations, cardiac surgical techniques	Counseling