Cardiovascular Conditions

Overview of Congenital Heart and Valve Diseases

**Definition**

- **Congenital heart disease:** Anatomic defects of the heart and great vessels present at birth

The incidence is 1/120 live births. Some common causes are chromosomal defects (eg, trisomy 13 or 18), maternal illness (eg, diabetes mellitus, fetal alcohol syndrome, rubella), medication (eg, thalidomide).

- **Mitral valve disease:** A bulging of one or both mitral valve leaflets into the left atrium during systole

Between 1% and 6% in otherwise normal populations. It is higher in persons with Duchenne MD, myotonic dystrophy, sickle cell disease, atrial septal defect, and rheumatic heart disease. About 25% of patients have joint laxity, a high-arched palate, or other skeletal abnormalities.

- **Aortic valve disease:** Retrograde flow from the aorta into the LV through incompetent aortic cusps

Incidences of aortic regurgitation usually increase with age. Common causes are congenital (eg, born with a bicuspid valve), idiopathic degeneration of the aortic valves or root, rheumatic heart disease, infective endocarditis, and trauma. Less common causes are severe hypertension and some autoimmune diseases.

- **Tricuspid valve disease:** Retrograde flow of blood from right ventricle to right atrium due to inadequate apposition of the tricuspid valves

Due to a cleft tricuspid valve (eg, in endocardial cushion defects), blunt trauma, or carcinoid disease, in which the valve may be fixed in a semi-open position. Less common causes are infective endocarditis, papillary muscle dysfunction, right ventricle infarction, or the use of fenfluramine.

**Prevalence**

- **Between 1% and 6% in otherwise normal populations.**

**Etiology and Pathophysiology**

- Many congenital cardiac defects do not produce significant hemodynamic alterations. Others cause abnormal ventricular volume load; ventricular pressure load; and atrial emptying, venous admixture, or inadequate systemic CO.

- Complete myxomatous degeneration of the valve can lead to severe mitral regurgitation or floppy valve syndrome.

- LV volume and LV SV are increased because the LV receives blood regurgitated during diastole in addition to normal blood flow from the pulmonary veins. LV hypertrophy occurs proportionally with dilation in order to maintain pressure.

- Severe pulmonary hypertension or right ventricle outflow obstruction leads to right ventricle dilation frequently, which in turn can cause tricuspid valve regurgitation.

- Fatigue, cold skin, dyspnea, edema, and the sensation of pulsations in the neck due to the high jugular regurgitant are common. Right upper quadrant abdominal discomfort due to hepatic congestion may occur. AF or flutter, which usually occurs when the right atrium enlarges, further decreases the CO and may precipitate sudden, severe HF.

**Clinical Presentation and Course**

- Heart murmurs due to turbulent flow are common. Signs of HF, cyanosis, and hepatomegaly may be present in the newborn. Long-standing hypoxemia can lead to clubbing, polycythemia, and other signs of inadequate systemic perfusion. Dilation and hypertrophy of cardiac chambers may result from increased cardiac workload.

- In mild cases, patients are asymptomatic. Patients might have a crisp systolic sound or click and a delayed or late systolic mitral regurgitation murmur. In more severe patients, arrhythmias, palpitations, syncope, fatigue, light-headedness, TIA, dyspnea, and hemoptysis and abnormal EKG findings despite having normal coronary angiograms.

- Dyspnea on exertion, orthopnea, and paroxysmal nocturnal dyspnea and palpitations may occur. Angina is especially common at night.

Abbreviations: CO, cardiac output; MD, muscular dystrophy; TIA, transient ischemic attack.