N5315 Advanced Pathophysiology
Congenital Heart Defects

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Introduction

CHD are the most common heart disease which affect children. Preterm babies are more commonly affected than full term babies. Unfortunately, the etiology is unknown in most of the cases (90%). The known causes include genetic environmental interactions (multifactorial factors), primary genetic factors (single gene disorders, chromosome disorders) and sole environmental factors (isotretinoin, alcohol, maternal rubella infection).

Maternal risk factors include: age > 45, a prior child born with CHD, poor controlled diabetes during pregnancy, the use of alcohol during pregnancy, congenital infection during pregnancy (rubella), aspirin intake, systemic lupus, diphenylhydantoin (phenytoin) intake.

Left-sided to right-sided heart shunts

In a left to right shunt oxygenated blood from the left side of the heart mixes with the right side of the heart. The SaO_2 on the left side of the heart is 95% and normally on the right side it is 75%. As the blood mixes, the SaO_2 on the right side of the heart is increased to 80% or more.

The pathological consequences of a left to right shunt are directly caused by the amount of blood being shunted. Volume overload occurs in the right side of the heart. This leads to pulmonary hypertension, right ventricular hypertrophy (secondary to pulmonary HTN), left ventricular hypertrophy (secondary to increased blood return to the LV), and later a reversal of the shunt because the right side of the heart will end up with higher pressures than the left and the shunt reverses. This is known as Eisenmenger syndrome. This is considered late onset cyanosis and will result in clubbing of the fingers.

Ventricular septal defects are the most common CHD. This results in a communication (a hole) in the interventricular septum. This allows blood to move from the left ventricle into the right ventricle. Multiple VSDs are associated with tetralogy of Fallot and other congenital disorders such as Cri du chat syndrome, fetal alcohol syndrome. VSD is also associated with the development of atrial septal defects, patent ductus arteriosus, coarctation of the aorta, aortic valve stenosis. The infant/child will have a harsh holosytolic murmur at the lower, left sternal border. They are at an increased risk of developing endocarditis at some point in their lifetime.

Atrial Septal Defect is the most common CHD in adults. The most common cause is a patent foramen ovale that does not close. ASD is associated with fetal alcohol syndrome and Down syndrome. They will have a soft midsystolic murmur at the upper sternal border. They are at an increased risk of developing an embolus.

Patent Ductus Arteriosus defects results from a PDA which does not close after birth. PDA is associated with congenital rubella, respiratory distress syndrome after birth, complete transposition of the great vessels, and tetraology of Fallot. A machine like murmur is heard continuously through systole and diastole. A reversal of the shunt may occur if pulmonary

hypertension develops from increased blood flow through the pulmonary artery. If this happens deoxygenated blood will enter the aorta below the level of the subclavian artery. The child will be cyanotic in the lower body but normal in the upper body

Right- to left-sided heart shunts

In a right to left shunt deoxygenated blood is mixed with oxygenated blood. The SaO_2 on the left side of the heart drops from 95% to 80% or lower. The development of cyanosis is dependent on how low the SaO_2 is on the left side of the heart. If the SaO_2 remains at 85% or higher cyanosis is not likely to develop because the shunting is minimal. An SaO_2 below 80% is indicative of a higher amount of blood being shunted and will result in cyanosis. Tetralogy of Fallot is the most common cyanotic CHD.