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CONGENITAL HEART DISEASES IN NEWBORNS : EARLY DETECTION AND OUTCOMES

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Introduction: Development of the infant heart (Embryology)

1. Formation of the primitive heart tube it starts from week 3 of gestation period. It's origin is from splanchnic mesoderm where the two endocardial tube fuse together to form a single primitive heart tube.There are 5 segments which is from cranial to caudal : Truncus arteriosus,bulbus cordis,primitive ventricle,primitive atrium,sinus venosus.If any deviation occurs in these segments it can lead to complex congenital heart disease (CHD).

2. Cardiac looping happens from week 4 of the gestation period. In this case the heart tube undergoes rightward looping (D-looping).This formation matters because it develops a normal spatial arrangement.The right ventricle comes to lie in the front ie anterior.The left ventricle moves to the back ie posterior. If the looping is not normal it can cause two abnormal looping in heart ie Dextrocardia - heart is located on the right side instead of left and congenitally corrected transposition of the great arteries - abnormal arrangement of the major heart vessels.

3. Formation of the heart septa starts from weeks 4-8 of the gestation period.Septation of is the process by which the early heart(a single tube) is divided into four chambers → right atrium,left atrium,right ventricle and left ventricle.There will be formation of the atrial septa,ventricular septa and atrioventricular canal division. Atrial septum forms → divides the

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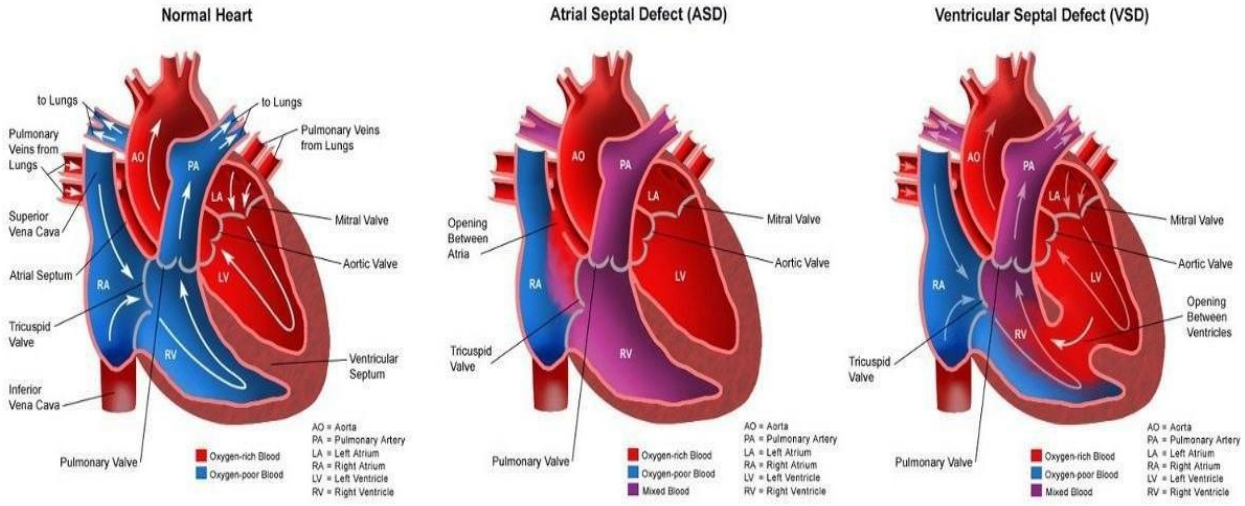

upper chambers(ventricles). Ventricular septum forms → divides the lower chambers(ventricles).

Atrioventricular canal divison where the endocardial cushions divide AV canal into Mitral and Tricuspid valve. Important because it ensures proper separation of oxygenated and deoxygenated blood also allows the heart to function efficiently after birth. At birth the atrial septation is functionally closure due to the increase in left atrial pressure.If there is abnormality in septal formation it can lead to congenital heart defects such as atrial septal and ventricular septal defect.

4. Conotruncal septation (Aorticopulmanory) is a process by which the single outflow tract of the primitive heart is divided into two major arteries. These are aorta (which carries the oxygenated blood to the body) and pulmonary artery (which carries deoxygenated blood to the lungs). There are special cells called neural crest cells migrate from the developing nervous system into the heart and these cells from a spiral shaped wall (septum) inside the outflow tract which is known as Aorticopulmanory septum if this process fails it can lead to the failure of the neural crest migration of septum formation causes serious congenital heart diseases.

5. Development of valves is a complex process that occurs during the fourth through eight weeks of embryonic development. It is essentially transforms the heart from a simple tube into a multi -chambered pump with one way gates.

Atrioventricular valve:- the Mitral and Tricuspid valves which is separating the atrial from the ventricles.

* The role of endocardial cushions:These are thickenings of specialized tissues that grow from the walls of the heart towards the center.As they fuse,they divide the single atrioventricular canal into the right and left openings.

* Formation through excavation: After the canal is divided, the surrounding tissue undergoes a process of remodeling. The ventricular walls excavate or hollow out,leaving behind thin strands of tissue.

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* The final structure: These strands eventually differentiate into the valve leaflets (cusps),the chordae tendineae and the papillary muscle that anchor the valves.

Semilunar valves:- The aortic and pulmonary are located at the exists of the heart, where blood leaves the ventricles to enter the great arteries.

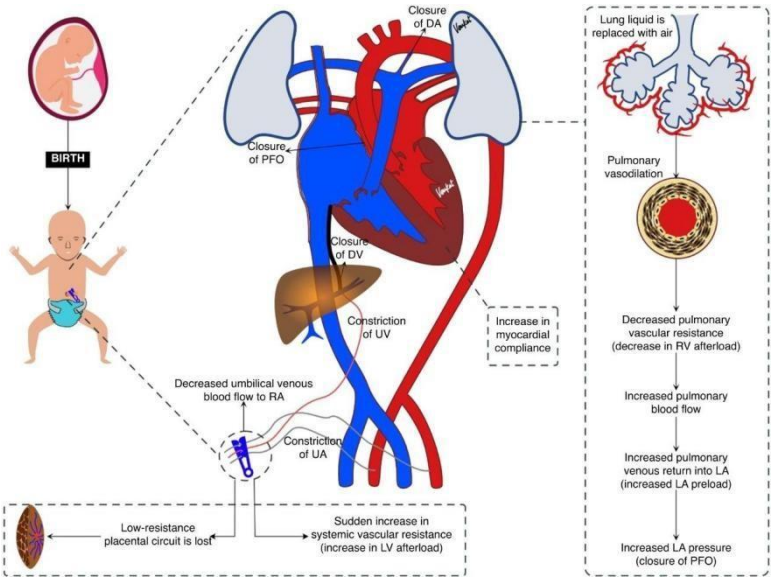
* The truncal swellings: This valves develop from small masses of tissue called truncal swelling or ridge located within the Truncus arteriosus that is the single large vessel that eventually splits into the aorta and pulmonary artery.

* Division and sculpting: As the Truncus arteriosus is divided into two separate vessels by the spiral septum, these swellings are rearranged.

Three swellings are assigned to each new vessel.

* Formation of the cusps: The tissue is “hollow out” from the top down,creating three thin, cup-shaped pockets. Because they look like half- moon,they are called semilunar.

Fetal Circulation :- In a fetus,the lungs and the liver are not fully functional,so the body uses special shunt (bypass pathway) to circulate blood efficiently. There are some key shunts as follows:



✓ Foramen ovale :

* An opening between the right and left atrium.

* Allows oxygenated blood from the placenta to bypass the lungs and go directly into systemic circulation.

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* Blood flow:Right atrium- Left atrium- left ventricle- aorta.

✓ Ductus arteriosus:

* A vessel connecting the pulmonary artery to the aorta.

* Diverts blood away from the lungs.

* Blood flow: pulmonary artery - aorta.

✓ Ductus venosus :

* connects the umbilical vein to the inferior vena cava (IVC).

* Allows blood to bypass the liver and flow directly into circulation.

7. Transition at birth:- when the baby takes the first breath, major changes occur:

✓ Lungs expand:

* Air enters lungs- Pulmonary vessels open .

* Blood now flows to lungs for oxygenation.

✓ Pressure changes in heart:

* Left atrial pressure becomes higher than right.

* This causes closure of the foramen ovale.

✓ Closure of shunts:

* Foramen ovale - becomes fossa ovalis.

* Ductus arteriosus - becomes ligamentum arteriosum.

* Ductus venosus - becomes ligamentum venosum.

✓ Umbilical cord is cut:

* Placental circulation stops.

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* Baby becomes independent for oxygen and nutrients.

> Fetal Circulation: The fetus means baby in the womb gets oxygen and nutrients from the mother through the placenta because it's lungs are not working yet. There are following steps of blood flow in the infants :

1. The oxygenated blood comes from the placenta through the umbilical vein.
2. It passes through the liver, mostly bypassing it via the ductus venosus, and enters the interior vena cava.
3. Blood enters the right atrium of the heart.
4. Most of it passes directly to the left atrium through the foramen ovale(bypassing the lungs)
5. From the left atrium → left ventricle → pumped into the aorta → supplies the body.

What happens to blood when it goes to the lungs:

• some blood from the right atrium goes to the right ventricle → pulmonary artery. Since lungs are not functional most blood is diverted to the aorta through the ductus arteriosus.

There are fetal structures which includes :

One Umbilical vein which brings deoxygenated blood.
ductus venosus which bypass the liver. foramen ovale which connects the right and left atria.
ductus arteriosus which connects the pulmonary artery to aorta.
Two Umbilical arteries which carry deoxygenated blood to placenta.

✓ Congenital Malformation Syndrome and Cardiac Features

1. Chromosomal abnormalities: Trisomy 21 (down Syndrome)

* causes: Human cells typically contain 23 pairs of chromosomes. Trisomy 21 occurs due to an error in cell division called nondisjunction.

NONDISJUNCTION:

This is the happen when a pair of chromosomes fails to

separate properly during the formation of an egg or sperm cell. When that cell joints with normal cell the resulting embryo has three copies of chromosomes 21 instead of two.

VARIATIONS: STANDARD TRISOMY 21(95%):

Every cell in the body has three copies of chromosomes 21.

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. MOSAICISM: only some cells have the extra chromosomes;
symptoms may be milder.

. TRANSLOCATION: part of chromosomes 21 becomes attached
to another chromosomes.

CONSEQUENCE AND FEATURES:

The extra genetic material affect the development of various
body system .

. Physical Features: common traits includes a flattened facial profile,upward slanting eyes (epicanthal folds) , single deep crease across the palm.

INTELLECTUAL AND DEVELOPMENTAL:

Most individual have mild to moderate intellectual disability
and delays in speech and motor skills.

MARFAN SYNDROME IN NEWBORNS (NEONATAL MARFAN SYNDROME):

It's a genetic disorder of connective tissue that affects the heart, blood vessels, eyes , and skeleton.

It is caused by mutation affecting the body structural proteins, making tissue weaker and more elastic than normal.

CAUSES:

. Mutation in the FBN1 gene.
. This gene produce fibrilline -1.

. Usually autosomal dominant inheritance
. In some newborns, it occurs due to new (spontaneous) mutation.

CARDIAC PROBLEMS IN NEWBORN

MARFAN syndrome is strongly linked to congenital and early onset heart diseases:

Common heart issues:

.Mitral valve prolapse → valve doesn't close properly

.Severe Mitral or Tricuspid regurgitation

.Enlargement of the aorta (Aortic aneurysm)

.Risk of aortic dissection (life-threatening)

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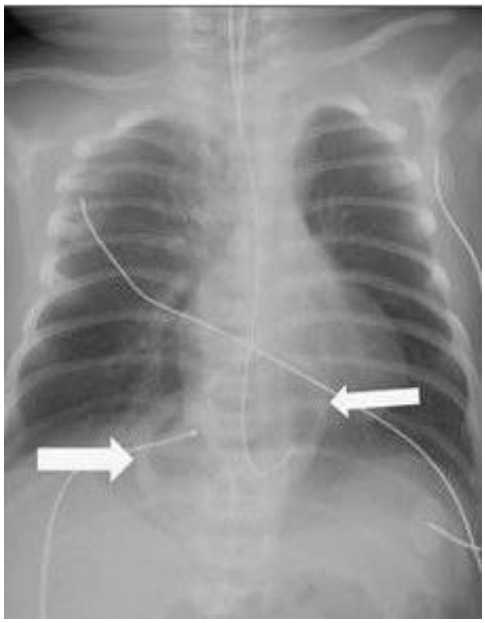
SIGNS AND SYMPTOMS IN NEWBORNS

Long limbs,fingers,loose joints,poor feeding and failure to

thrive,rapid breathing or difficulty breathing,heart murmur,enlarged heart,signs of heart failure.

DIAGNOSIS :- clinical examination (doctor look for physical features and heart signs) ,imaging (echocardiography → main tool to detect valve problems and aortic enlargement) , genetic testing (confirm mutation in the FBN1 gene ,diagnostic criteria (based on the Ghent Criteria).



> Conclusions : Congenital heart disease in newborn are a major cause of illness and death World wide. Early diagnosis and proper treatment are very important. These conditions may occur due to structural defect in the heart of genetic disorders such as MARFAN syndrome, affecting the normal function of the cardio vascular system from birth.

With modern diagnostic tools, especially echocardiography, early detection has become easier and more accurate. This helps doctors start treatment on time, whether medical or surgical. Advance in treatment matters, including drugs and corrective surgeries, have greatly improved survival rates and long term outcomes.

However, continuous follow up and a team-based approach to care are still essential. Regular monitoring helps prevent complications and supports proper growth and development. Improving newborn screening, increasing awareness, and ensuring access to specialized cardiac care are important

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