

In Syndrome

Ebstein's Anomaly

Definition :

- It is a congenital abnormality & the basic defect in this is tricuspid leaflets are displaced into the right ventricle.
- Both the tricuspid valve and the right ventricle are dysplastic, with it frequently described as "atrialization" of the right ventricle.

History :

- It was first described by the pathologist Wilhelm Ebstein in 1866 when he performed an autopsy of a laborer who suffered from exertional dyspnea all his life, had evidence of cyanosis and died of a sudden cardiac arrest.

Etiology :

- It is associated with chromosome 15q, which is involved in the embryological development of the heart in utero.
- Also a possible association of Ebstein anomaly with chromosome 11q.
- Studies suggest it may be associated with mutations in myosin heavy chain 7 (frequently associated with cardiomyopathies) and NKX2.5 (associated with other congenital heart defects).
- The teratogenic effect of Lithium.

Epidemiology :

- 0.3% to 0.6% of all congenital heart defects.
- 0.2 to 0.7 per 10,000 live births.

Clinical Features : In neonates :-

- Cyanosis
- Tricuspid regurgitation

Right ventricular dysfunction In early childhood :-

- A prominent v wave in jugular venous pulsation due to severe tricuspid regurgitation
- A loud first heart sound due to anterior leaflet closure, referred to as the "sail sound," and the first heart sound can also be split due to delayed tricuspid valve closure.

- A holosystolic murmur of tricuspid regurgitation is another common physical examination finding in these patients.
- Multiple ejection clicks can be heard in the case of a very mobile anterior leaflet.

In adults :-

- Palpitations
- Exertional dyspnea
- Chronic fatigue
- Lower extremity edema

Diagnosis :

- **X ray chest** – Globular heart, enlarged right atrium and oligemic lung fields.
- **ECG** – RBBB with prolonged PR interval, large P waves (Himalayan P waves), indicating right atrial enlargement in 10% cases WPW syndrome may occur.
- **Echo** – It confirms the anomaly. There is abnormal positional relationship between Mitral & Tricuspid valves with septal displacement of the septal tricuspid leaflet.

Treatment :

- **Medical:-** Treat Right sided heart failure.
- **Surgical :-**
 - ✓ Tricuspid valve replacement and closure of ASD.
 - ✓ Tricuspid Annuloplasty with placcation of the atrialized portion of RV.
- **Indication for surgery :**
 - ✓ Severe decompensation of heart.
 - ✓ Severe tricuspid regurgitation.
 - ✓ Cardiothoracic ratio > 60%
 - ✓ Patient having cyanosis due to foramen ovale or presence of ASD.

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