Ebstein’s Anomaly

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Agenda

• Ebstein’s Anomaly:
  – Introduction
  – Tricuspid valve anatomy and development
  – Pathophysiology
  – Epidemiology

• Patient Cases:
  – In utero diagnosis
  – Neonatal presentation
  – Childhood/Adult diagnosis
Ebstein’s Anomaly: Introduction

• First described in 1866 by German physician Dr. Wilhelm Ebstein:
  – Young man with palpitations and shortness of breath, found on autopsy to have abnormalities of his right ventricle and tricuspid valve

• The Problem: Apical displacement of tricuspid valve leaflets

• 1/20,000 live births

• Association with maternal lithium use in early pregnancy

Tricuspid Valve Development

- Normal tricuspid valve has 3 leaflets:
  - Septal
  - Anterior
  - Posterior

- Leaflets form by separating from the myocardium, or delamination
- Ebstein’s anomaly is caused by a failure of delamination, resulting in tethering of the leaflet to the myocardium
- The free edge of the leaflet is displaced apically

Modified from: Lacro, et al., Atlas of Heart Disease, 2002
Ebstein’s Anomaly: Pathophysiology

1. Failure of delamination of septal and posterior valve leaflets
2. Apical displacement of functional tricuspid valve
3. RV with 2 components: atrialized (ARV) and functional RV (RV)
4. Tricuspid regurgitation with RA and RV dilation -> progression to CHF
5. **Cyanosis:** if associated with a PFO or ASD, causing R->L shunt

Image source: Sridharan, et al., “Ebstein’s Anomaly”, Cardiovascular MRI in Congenital Heart Disease, 2010
Ebstein’s Anomaly: Pathology

- Dilated right atrium
- Dilated right ventricle
- Small functional right ventricle
- Bowing of IV Septum
- Posterior Leaflet

Ebstein’s Anomaly: Clinical Presentation

- Range of severity, depending on extent of tricuspid valve displacement

- Can present at any age:
  - Fetuses: Abnormal fetal screening ultrasound
  - Neonates: Cyanosis and hypoxia
  - Infants: Heart failure (poor feeding, lethargy, failure to thrive)
  - Children: Incidental murmur
  - Adolescents and adults: Arrhythmia, exertional dyspnea

- Often associated with other congenital heart defects: PFO/ASD, VSD, pulmonary stenosis, pulmonary atresia, PDA, and coarctation are most common

- Associated with WPW and arrhythmias due to presence of accessory pathways around malformed tricuspid valve
Patient 1: Pregnant Mother

28 year old G1P0 healthy female with an uncomplicated pregnancy comes to the office for her 18-week fetal survey
Patient 1: OB Ultrasound

**Normal Transabdominal Ultrasound**
(for comparison)
Axial View of Fetus

**Patient 1: Abnormal cardiac findings**
Transabdominal Ultrasound
Axial View of Fetus

- Uterus
- Spine
- Ribs
- Lung
- Cardiomegaly
- Dilated right atrium
- Horizontal rotation of heart
Patient 1: Pregnant Mother

Given the abnormal fetal cardiac findings on ultrasound, our patient was referred for a fetal echocardiogram.
Fetal Echocardiography

• Detailed ultrasonographic assessment of fetal cardiac structure and function

• Usually performed between 18-22 weeks

• Can be performed transabdominally or transvaginally

• Indications:
  – Maternal: family history of CHD, diabetes, autoimmune disorders, teratogen exposure
  – Fetal: abnormal cardiac screening exam, abnormal HR, chromosomal anomaly, extracardiac anomaly

• Preferred over fetal MRI:
  – Motion artifact: fast fetal heart rate and fetal movements
Patient 1, 29 weeks: Fetal Echocardiogram

Four-Chamber View

- RA
- ARV
- LA
- Ao
- RV
- LV

- Spine
- Ribs
- Lung
- PFO

Dilated RA and ARV
Apical displacement of the tricuspid valve
Patient 1, 29 weeks: Fetal Echocardiogram

Four-Chamber View, Doppler

Tricuspid Regurgitation
Summary: In utero diagnosis of Ebstein’s Anomaly

- Imaging modalities:
  - Fetal Ultrasound
  - Fetal Echocardiography

- Our patient’s fetal echocardiographic findings were diagnostic of Ebstein’s Anomaly:
  - Apical displacement of tricuspid valve leaflets
  - Right atrial dilation and severe cardiomegaly
  - Tricuspid regurgitation

- Diagnosis can be made as early as 14-16 weeks gestation at end of embryogenesis

- In utero diagnosis depends on severity of disease
Patient 2: Newborn Boy

**HPI:** Baby boy (BB) born to Patient 1 at 37 + 2/7 days by forceps-assisted vaginal delivery. APGARS 8 and 8, good tone, breathing spontaneously but poor respiratory effort.

**Physical Exam:**

HR 60, O2sat 50%

Cyanotic, 3/6 systolic murmur, lungs clear, good distal pulses and capillary refill < 3sec

BB was intubated and transferred to the Cardiac Intensive Care Unit. A CXR was done on day of life 1.
Patient 2: Chest X-Ray

- Enlarged cardiothymic silhouette
- Small amount of aerated lung visible at the right costaphrenic sulcus
- Endotracheal tube
- Umbilical vein catheter

Image source: Alvares et al., *Radiologia Brasileria*, 2006

Patient 2: AP Supine CXR

Normal AP Supine CXR (for comparison)
Ebstein’s Anomaly: “Box-Shaped” Heart

- Enlarged right atrium and right ventricle
- Pulmonary trunk obscured by enlarged RV giving “squared-off” cardiac contour
- Filling of retrosternal space by enlarged RA
- Decreased pulmonary markings (not seen in this CXR)
Summary: Chest X-Ray Findings in Ebstein’s Anomaly

• Key Findings on CXR:
  – Enlarged cardiac silhouette
  – “Box-shaped” heart
  – Decreased pulmonary markings

• Non-specific

• Good initial test, but a normal CXR cannot rule out Ebstein’s anomaly
Patient 2: Echocardiogram
Four-Chamber View

Dilated RA and ARV
Apical displacement of the tricuspid valve
PFO

Play Cine
Ebstein’s Anomaly: Leaflet “Tethering”

In Ebstein’s Anomaly, the tricuspid valve leaflets are “tethered” to the myocardium, resulting in apical displacement of the valve.


Companion Patient 2: Four-chamber Echocardiogram
Patient 2: Echocardiogram
Four-Chamber View, Doppler: Tricuspid Regurgitation
Patient 2: Echocardiogram 3D Reconstruction

A) Long-axis view of the right heart. **Apical displacement** of the tricuspid valve and **dilated RA and ARV** are noted.

B and C) Short-axis view of the tricuspid valve, with RV projecting out of the screen and RA projecting into the screen, in **diastole (B)** and **systole (C)**. **Noncoaptation** of the tricuspid valve leaflets is noted in systole, causing tricuspid regurgitation.

Children’s Hospital Boston, Courtesy of Dr. Vassilios Bezzerides
Summary: Echocardiography

• Diagnostic test of choice

• Pros:
  – Detailed assessment of cardiac structure and function
  – No contraindications, relatively easy
  – Can assess severity of regurgitation and dilation
  – Measure RV function
  – Identify any associated cardiovascular anomalies

• Cons:
  – Can be inconclusive
  – Operator-dependent

• GOSE: Great Ormond Street Score for neonates
  – Ratio of RA + ARV size to combined area of functional RV, LA, LV
  – Greater ratio = worse prognosis
Patient 3

**HPI:** 64yoF smoker with HTN and a history of an unknown “congenital heart disease” who presents with worsening exertional dyspnea and fatigue. No chest pain, pedal edema, orthopnea, or palpitations.

**Physical Exam:**
- Afebrile, HR 60, BP 140/80
- Notable only for 3/6 systolic murmur

**Echo:** “Technically difficult” study with suboptimal views. Showed markedly enlarged RA and RV, linear mid-RV band causing tricuspid regurgitation.

An MRI was obtained.
Cardiac MRI

• Same principles as MRI, but optimized for viewing the heart:
  – Different sequences to assess: function, morphology, perfusion, viability/infarction, flow, and angiography
  – EKG Gating: Acquire images at each stage of cardiac cycle of several beats

• Indications:
  – Non-diagnostic echocardiogram
  – Surgical planning

• Cons:
  – Expensive
  – Contraindicated in patients with claustrophobia, metallic hardware (including pacemakers*)

* Patients with Ebstein’s anomaly are at increased risk for arrhythmias, including WPW and other tachyarrhythmias due to accessory pathway around malformed tricuspid valve.
Patient 3: Cardiac MRI

Apical displacement of septal leaflet of tricuspid valve

Normal location of mitral valve

RA and RV Dilation

Axial MRI, + gadolinium
Patient 3: Cardiac MRI

Axial MRI, + gadolinium, CINE
Patient 3: Cardiac MRI

Coronal MRI, + gadolinium

Sagittal MRI, + gadolinium

Cardiomegaly

Dilated RV
Summary: Ebstein’s Anomaly

“Apical displacement of septal leaflet of tricuspid valve”

• Range of severity

• Can present at any age:
  – Fetuses – abnormal routine screening US
  – Neonates – cyanosis and hypoxia
  – Infants – heart failure
  – Children, adolescents, adults – incidental murmur, exertional dyspnea, arrhythmias

• Key Imaging Findings:
  – Cardiomegaly (from enlarged RA and RV) and “box-shaped heart”
  – Decreased pulmonary markings
  – Apical displacement of tricuspid valve
  – Tricuspid regurgitation
Summary: Imaging Modalities

1. CXR
   - Initial test of choice
   - Findings are nonspecific
   - Normal CXR does not rule out diagnosis

2. Echocardiography
   - Diagnostic test of choice
   - Assess severity of lesion and quantify regurgitation
   - Assess cardiac function

3. Cardiac MRI
   - Depiction of anatomy and function with unrestricted field of view
   - Imaging of pulmonary arteries
   - Useful when Echo is non-diagnostic

(4. Cardiac Catheterization)
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References


