EBSTEIN ANOMALY

Author Name: Mrs. Jovita Annie Alex

Assistant Lecturer

K. J. Somaiya College of Nursing

Introduction

Ebstein anomaly is a congenital malformation of the heart that is characterized by apical displacement of the septal and posterior tricuspid valve leaflets, leading to atrialization of the right ventricle with a variable degree of malformation and displacement of the anterior leaflet.

Wilhelm Ebstein first described a patient with cardiac defects typical of Ebstein anomaly in 1866. In 1927, Alfred Arnstein suggested the name Ebstein's anomaly for these defects. In 1937, Yates and Shapiro described the first case of the anomaly with associated radiographic and electrocardiographic data

Definition

Ebstein anomaly is a congenital malformation of the heart that is characterized by apical displacement of the septal and posterior tricuspid valve leaflets, leading to atrialization of the right ventricle with a variable degree of malformation and displacement of the anterior leaflet.

Epidemiology

Ebstein anomaly probably accounts for 0.5% of cases of congenital heart diseases. Its true prevalence is unknown because mild forms frequently are undiagnosed. With the wide application of echocardiography, more cases are being diagnosed.

Race-, sex-, and age-related demographics

Ebstein anomaly is more common in children of white females. However, no specific sex predominance exists.

Ebstein anomaly can present at various stages of life, as follows:

- Fetal life: Ebstein anomaly is usually diagnosed incidentally by echocardiography.
- Neonatal life and infancy: Ebstein anomaly presents with cyanosis and/or severe heart failure; typically, symptoms present in infancy improve as pulmonary vascular resistance decreases.

• Adult life: Ebstein anomaly presents with fatigue, exertional dyspnea, cyanosis, tricuspid regurgitation and/or right heart failure, and palpitations; arrhythmias are common.

Etiology

Ebstein anomaly is a congenital disease of often uncertain cause.

Environmental factors implicated in etiology include the following maternal factors :

- Maternal ingestion of lithium in first trimester of pregnancy
- Maternal benzodiazepine use
- Maternal exposure to varnishing substances
- Maternal history of previous fetal loss

The risk of having Ebstein anomaly is higher in white persons than in other races.

Pathophysiology

The embryological development of tricuspid valve leaflets and chordae involves undermining of the right ventricular free wall. This process continues to the level of the atrioventricular (AV) junction. In Ebstein anomaly, this process of undermining is incomplete and falls short of reaching the level of the AV junction. In addition, the apical portions of the valve tissue, which normally undergo resorption, fail to resorb completely. This results in distortion and displacement of the tricuspid valve leaflets, and a part of the right ventricle becomes atrialized. In one study involving 50 hearts with the anomaly, the entire right ventricle was found to be morphologically abnormal.

Ebstein anomaly is commonly associated with other congenital, structural, or conduction system disease, including intracardiac shunts, valvular lesions, and accessory conduction pathways (eg, Wolff-Parkinson-White [WPW] syndrome).

The hemodynamic consequences of Ebstein anomaly result from displaced and malformed tricuspid leaflets and atrialization of the right ventricle. The leaflet anomaly leads to tricuspid regurgitation. The severity of regurgitation depends on the extent of leaflet displacement, ranging from mild regurgitation with minimally displaced tricuspid leaflets to severe regurgitation with extreme displacement.

The atrialized portion of the right ventricle, although anatomically part of the right atrium, contracts and relaxes with the right ventricle. This discordant contraction leads to stagnation of blood in the right atrium. During ventricular systole, the atrialized part of the right ventricle contracts with the rest of the right ventricle, which causes a backward flow of blood into the right atrium, accentuating the effects of tricuspid regurgitation.

Clinical Manifestations

Patients can have a variety of symptoms related to the anatomic abnormalities of Ebstein anomaly and their hemodynamic effects or associated structural and conduction system disease, including the following:

- Cyanosis- is fairly common and frequently due to right-to-left shunt at the atrial level and/or severe heart failure. It is transient in neonatal life, with recurrence in adult life; it may appear for the first time in adult life. The transient appearance/worsening of cyanosis in adult life is due to paroxysmal arrhythmias; once apparent, the cyanosis progressively worsens.

- Fatigue and dyspnea

- Palpitations and sudden cardiac death- may occur due to paroxysmal supraventricular tachycardia (SVT) or Wolff-Parkinson-White (WPW) syndrome in as many as one third of patients. Fatal ventricular arrhythmias may also occur due to the presence of accessory pathways.

- Symptoms of right heart failure, such as edema and ascites

Other less common presenting symptoms include the following:

- Brain abscess due to right-to-left shunt

- Bacterial endocarditis

- Paradoxical embolism, stroke, and transient ischemic attacks

Diagnosis

Physical findings in patients with Ebstein anomaly span a spectrum from subtle to dramatic. They may include the following:

- Cyanosis and clubbing

- Precordial asymmetry

- Jugular venous pulse

- Arterial pulses: Usually normal but are diminished late in the disease course

- Heart sounds: Widely split first heart sound with loud tricuspid component and soft/absent mitral component in the presence of prolonged PR interval; usually normal second heart sound but may be widely split when pulmonary component delayed due to right bundle-branch block; third

- and fourth heart sounds commonly present, even in the absence of congestive heart failure

Testing

A 12-lead electrocardiogram may demonstrate the following findings in patients with Ebstein anomaly:

- Normal sinus rhythm

- Intermittent/paroxysmal supraventricular tachycardia, atrial flutter/fibrillation, ventricular tachycardia
- Abnormal P waves consistent with right atrial enlargement
- Prolonged PR interval; may be normal/short in patients with Wolff-Parkinson-White syndrome
- Right bundle branch block and/or low voltage QRS complex

Imaging studies

The following imaging studies may be used to assess patients with suspected Ebstein anomaly:

- Chest radiography
- Echocardiography: Includes M- mode, 2-dimensional, and Doppler studies
- Cine magnetic resonance imaging: In selected patients; not used routinely

Management

Treatment of Ebstein anomaly is complex and dictated mainly by the severity of the disease itself and the effect of accompanying congenital structural and electrical abnormalities. Treatment options include medical therapy, radiofrequency ablation, and surgical therapy.

Pharmacotherapy

Ebstein anomaly requires drug treatment for cardiovascular consequences resulting from tricuspid atrialization of the right ventricle, valvular regurgitation, and septal defects. Patients may require antibiotic prophylaxis for bacterial endocarditis.

The following medications are used in patients with Ebstein anomaly:

- Diuretics (eg, furosemide)
- Cardiac glycosides (eg, digoxin)
- Angiotensin-converting enzyme inhibitors (eg, enalapril)

Nonpharmacotherapy

Radiofrequency ablation of the accessory pathways is an alternative to medication for treatment of arrhythmias. Curative therapy of supraventricular tachycardia with radiofrequency ablation is the treatment of choice. However, in patients without significant structural heart disease, the success rate of this procedure is lower.

Surgical option

Surgical intervention includes the following:

- Correction of the underlying tricuspid valve and right ventricular abnormalities
- Correction of any associated intracardiac defects

- Palliative procedures in early days of life as a bridge to more definitive surgical treatment later
- Surgical treatment of associated arrhythmias (eg, ablation of the accessory pathways; maze procedure for atrial arrhythmias)
- Cardiac transplantation in selected patients

Prognosis

Prognosis depends on the severity of the disease and treatment options available. Pregnancy in women with Ebstein anomaly seems to be well tolerated with adequate supervision.

Poor prognostic signs include the following:

- Male sex
- Earlier age at presentation
- Cardiothoracic ratio of more than 0.65 on chest radiographs
- Septal leaflet attachment ratio (ie, ratio of distance between AV ring and distal attachment of septal leaflet to length of septal leaflet) of more than 0.45
- Increasing ratio of combined area of right atrium and atrialized right ventricle to that of the functional right ventricle—grade 1, which is less than 0.5, to grade 4, which is more than 1.5 (increase in the relative risk of 2.7 for each increment in the grade)
- New York Heart Association (NYHA) class: This has been linked with mortality rate in some studies

Morbidity/mortality

The natural course of the disease varies according to the severity of tricuspid valve displacement. Patients presenting in infancy generally have severe disease and unfavorable prognosis.

The mean age of presentation is in the middle teenage years. According to older observational data, approximately 5% of these patients survive beyond age 50 years. The oldest recorded patient lived to age 85 years.

Complications

Complications of Ebstein anomaly include the following:

- Congestive heart failure
- Sudden cardiac death
- Bacterial endocarditis
- Brain abscess
- Paradoxical embolism

- Transient ischemic attacks
- Stroke

Nursing Management

- Monitoring of vital the signs for a patient with Ebstein anomaly and the likely findings
- Life styles modification for the patient- caring environment which promotes rest for the patient in order to prevent the demand for oxygen
- Diet modification for the patient- golden rule of low salt, fat free formula, Vitamin rich food
- Administration of medications and oxygen therapy
- Coordination of the diagnostic examinations for efficient clinical management
- Provision of counseling and health education to the patient and the family
- Applications of ethical principles in caring for the patient with Ebstein anomaly

Case Study

A 24 year old G2A1 at 39 weeks 6 days gestation with a known case of Ebstein anomaly was referred to NEIGRIHMS on April 2017 for further management as our institute is having well equipped cardiac facilities. She was diagnosed as a case of cardiac anomaly during childhood and it was confirmed by ECHO during her treatment for infertility. All these years, she did not have any symptoms related to the condition and was not on any treatment. In the present pregnancy, she had regular antenatal checkups and the whole antenatal period was uneventful. On admission, the general condition of the patient was good. No pallor, icterus, cyanosis, oedema, clubbing was seen. On examination of the respiratory system, no abnormality was detected. Her pulse rate was 90 beats per minute, regular and blood pressure was 110/80 *mm Hg* and her jugular venous pressure was normal. Her oxygen saturation in room air was 98%. On cardiovascular system examination, a split S1 was heard with audible S2, S3 and S4. Pan systolic murmur was also heard. On ECG, right axis deviation with normal sinus rhythm was seen. Echocardiography showed apical displacement of septal leaflet of tricuspid valve, elongated anterior tricuspid leaflet, dilated right atrium and right ventricle, moderate tricuspid regurgitation, small ASD and normal right ventricular function with left ventricular ejection fraction of 60% . Echo findings confirmed diagnosis of Ebstein anomaly.

Blood group: O positive, haemoglobin 10.5 *gr/dl*, TLC and DLC: WNL, coagulation profile: normal, liver function tests and kidney function tests: normal, and RBS: 92 *mg/dl*. All viral markers were negative and thyroid profile was normal. Level II ultrasound obstetrics scan was done on the day of admission that showed single live intrauterine fetus with cephalic presentation; the gestational age was 37 weeks 4 days, placenta was fundo-body posterior, liquor was adequate, and expected fetal weight was 3.3 *kg*. Umbilical artery Doppler showed a normal study. No gross congenital anomaly in the fetus was observed. On per vaginal examination, her Bishop score was 4.

Cardiology opinion was taken and patient was advised to accept termination of pregnancy preferably by caesarean section to avoid prolonged induction of labor.

Elective LSCS was done at 40 weeks 3 days under epidural anaesthesia with antibiotic coverage (Injection of ampicillin 2 gram and injection of gentamycin 80 *mg* intravenously) for bacterial endocarditis prophylaxis. A healthy male baby weighing 2.5 *kg* was delivered. 10 units of oxytocin were given intramuscularly. Intraoperative and postoperative period was uneventful. After delivery, ECHO of baby was done, which was normal. Patient was discharged on the 7th postoperative day after suture removal with the advice to attend postnatal clinic and cardiology OPD after 1 month. On follow-up, patient was advised to use progesterone only pill to avoid pregnancy.

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