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Case Report

Paediatrics Section

Incidental Finding of Ebstein's Anomaly in an Adolescent with an Upper Respiratory Infection: A Case Report

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ABSTRACT

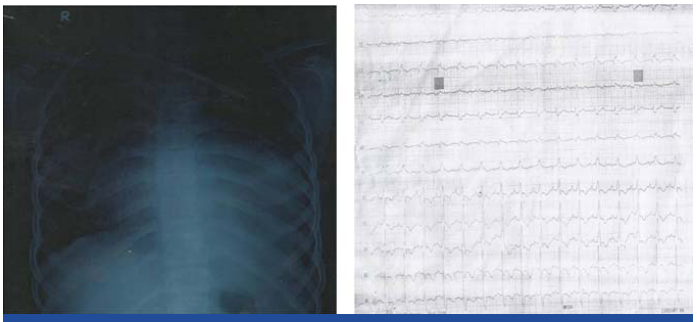
Ebstein's anomaly is a rare congenital heart disorder and has a varied clinical course, with detection as late as the seventh decade. We hereby describe an 11-year-old child in whom Ebstein's anomaly was diagnosed. The most common presentation in early second decade is due to an arrhythmia; however, the present case report is about a patient who presented with acute congestive heart failure due to decompensation from an acute upper respiratory tract infection, which is a rather uncommon presentation.

Keywords: Acute decompensated heart failure, Progressive dyspnea, Lack of arrhythmia

CASE REPORT

An 11-year-old Indian girl presented to an outpatient clinic with fever, cough and cold and progressive shortness of breath for 10 days. Her mother reported that she was always short of breath, first noticed when she was a toddler but did not seek any evaluation or treatment. Mother noticed that during these couple of days prior to admission, the child's dypnoea and purplish discoloration of lips had increased. She had a persistent, low grade fever and her cough was intermittent and increased at night. She denied chest pain. She was born to a non-consanguineous mother and father, requiring no delivery room interventions and had normal APGAR scores at birth. Mother was not receiving any medications in the antenatal period. Postnatal history was insignificant. There was no family history of connective tissue disorders in the family. She had a past medical history significant for intermittent palpitations, fatigue and bluish discoloration of lips on exertion. At presentation, she was febrile (38.2^{0}C), dyspneic, cyanotic, tachypneic and tachycardic with blood pressure at the 50% for height, age and gender. Saturations were 85% on room air in the right index finger. Weight of the child was between 15-25 percentile per (World Health Organization) standards; height was between 85-95 percentile and the BMI was between third and fifth percentile.

Head and neck examination revealed a normal fundus, pale inferior turbinates and rhinorrhea; she also had visible cyanosis of the oral mucosa and a grossly injected pharynx posterior to tonsillar pillars. She had a prominent precordium with a displaced apical impulse over the sixth intercostal space along the midclavicular line. Her first heart sound was widely split. There was an early systolic ejection murmur increasing during inspiration along with a gallop. Her jugular veins were distended with a prominent a wave and she had modified Ross class III paediatric heart failure. Her abdomen examination revealed hepatomegaly. However, the chest was clear on auscultation and had upper airway conducted sounds. Upper extremity examination revealed grade II clubbing of the fingers. The remainder of the physical examination was non-contributory. Complete blood count was within normal limits. The renal function tests, basic metabolic panel, liver function tests, and blood cultures were also normal. Arterial blood gases revealed renal compensation of respiratory alkalosis and an oxygen saturation of 85%. Her chest radiograph revealed cardiomegaly with cardiothoracic ratio of 0.75, right atrial enlargement and a boot shaped heart. Her electrocardiogram revealed a normal sinus rhythm and a regular ventricular rate of 100 beats/min with a QRS axis of 135 degrees. She had a prolonged PR interval and a normal QRS interval, an incomplete right bundle branch block (RBBB) and tall P waves with a T wave inversion in leads V2, V3 and V4 [Table/Fig-1]. The echocardiogram (Echo) revealed situs solitus, levocardia, dilated RA and RV, atrialization of right ventricle with a sail like anterior tricuspid septal leaflet, and a patent foramen ovale. Aortic valve and pulmonary valve morphology and Doppler were noted to be normal. The Great Ormond Street Echocardiogram (GOSE) score was 0.8 (grade II). The tricuspid annulus was found to be 51 mm (normal: 30-32 mm) and tricuspid regurgitation with downward displacement of septal leaflet of tricuspid valve with a displacement index of 13mm/m^2 was noted. Acute congestive heart failure was managed using digoxin and diuretics. She was given a loading dose of digoxin at 10mcg/kg over 24 hours and was subsequently started on a maintenance dose of 2 mcg/kg and put on oral tablets at the time of discharge. After acute management, she was managed surgically. Amiodarone was used to control ventricular and supraventricular tachyarrhythmias after surgical correction. She was put on 15 mg/kg/day orally once daily for two weeks followed by 5 mg/kg/day daily. The child was subsequently assessed and showed clinical improvement in her clinical status. Echocardiogram revealed a reduction in tricuspid valve diameter, and reduction of right ventricular diameter. Serial EKG recordings on follow-up visits did not reveal any arrhythmias.



[Table/Fig-1a,b]: 1a : Cardiomegaly with CT Ratio = 0.75, Right atrial enlargment, Boot shaped heart and Oligemic lung fields. 1b : EKG showing a sinus rhytm of 100/min, regular, with a QRS axis of 135 degree, increased PR interval and R'R pattern in V1, Tall p waves in V2, V3, V4, suggestive of Right atrial hypertrophy. This EKG also depicts 1st degree AV block.

DISCUSSION

Less than one percent of all congenital heart disease is attributed to Ebstein's anomaly (EA) [1]. Spectrum of EA ranges from a symptomatic neonate to an adult without symptoms. There have been case reports in fetuses, neonates, young children, adolescents and adults; only five percent of patients survive beyond the fifth decade [2,3]. The degree of displacement as well as the functional status of tricuspid leaflets determines the symptomatology. Thus,

patients with mild apical displacement of tricuspid leaflets remain asymptomatic; however, cases with abnormal leaflet attachment or displacement causing severe tricuspid regurgitation, along with severe right ventricular outflow tract obstruction, present early and result in death in utero or present as heart failure or cardiomegaly at birth or oedema in lungs or liver [4]. The most common presentation in younger age group is that of a life threatening arrhythmia [1]. EA can cause severe tricuspid regurgitation in utero, lead to heart failure, cardiomegaly, tachyarrhythmias, pulmonary hypoplasia and even hydrops fetalis [5,6]. This case report describes a patient with EA that was incidentally detected in an outpatient setting secondary to decompensation from an upper respiratory tract infection.

EA is characterized by either right ventricular endocardial adhesions to the tricuspid valve, anatomical tricuspid annulus dilation, anterior leaflet anomalies and apical displacement of septal and part of posterior valve leaflets, and/or atrialization of right ventricle [5,7]. The haemodynamic parameters in a typical case of EA depend upon the extent of displacement of tricuspid valve, the right atrial pressure, and the degree of right to left interatrial shunting and the size and function of right ventricular cavity. These parameters will also define the degree of cyanosis, arrhythmias and congestive heart failure, which are the typical presenting features [1,8]. Patients suffering from EA suffer from a myocardial disease known as Left Ventricular Non Compaction (LVNC) that is associated with gene on chromosome 14q12 known as MYH7, which is associated with a sponge like muscle tissue protruding into the left ventricle, leading to impaired contraction [9].

Arrhythmias in EA can be asymptomatic or symptomatic [10]. Adolescent patients most commonly present with a rhythm disturbance, an incidental murmur, cyanosis and/or heart failure [11]. Overall, approximately 14-20 percent of EA patients will have one or more accessory conduction pathways with Wolff-Parkinson-White syndrome [12]. This patient, contrary to the usual presentation, presented with the features of acute decompensated congestive heart failure in association with an upper respiratory tract infection in absence of arrhythmia. She, however, lacked the usual signs such as ascites, peripheral oedema which is usually seen in isolated tricuspid regurgitation [13]. Echo also ruled out unobstructed pulmonary blood flow which is generally seen in d-TGA with associated tricuspid atresia, which presents quite early.

For the diagnosis, two-dimensional echocardiography is a good initial test; however, it has less accuracy for calculating RV volume and ejection fraction [14]. However, the RV myocardial reserve, which is an important prognostic factor, cannot be predicted accurately by any of the above investigations.

Management of EA is observation, medical and/or surgical therapy. Commonly used medication in the cases presenting as AV reentrant tachycardia is procainamide. If the condition is associated with atrial fibrillation and pre-excitation, the agents include procainamide, flecainide, propafenone, dofetilide and ibutilide since these medications slow conduction in the accessory pathway. These medications should be considered before considering electrical cardioversion. However, our patient showed decompensation in the absence of an arrhythmia. Observation is advised for asymptomatic patients. If the infant is able to reach childhood without any symptoms, they usually do well for many years; thus, surgery could be postponed until the symptoms appear or begin to progress [15]. In rare cases, patients present with an atrial septal defect or patent foramen ovale, with mild or moderate tricuspid regurgitation; these patients may benefit from device closure to prevent the development of paradoxical emboli and improvement of cyanosis [1].

CONCLUSION

EA is a unique and interesting congenital heart disease with variable presentation. Thus, precise knowledge of the various presentations, timely diagnosis and appropriate management options are necessary. Thus, the physician should be cognizant that minor illnesses like upper respiratory infections can worsen cyanosis and may uncover underlying cardiac disease.

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