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TRILOGY OF FALLOT: AN UNUSUAL CLINICAL PRESENTATION

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FK concieved idea, did literature review, final drafting and reviewed case report. SF helped in acquiring photographs. All authors contributed significantly to the submitted manuscript.

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ABSTRACT

While dealing with major pathologies like in our case, most physicians are often familiar with typical features of disease but are not able to recognize the occasional atypical presentation of disease resulting in patient being under diagnosed or late diagnosed.

We present a case of 25 year old gentleman who initially presented with complains of 2 episodes of blood in vomiting and diagnosis of trilogy of Fallot was made due to evidence of typical findings on Trans oesophageal Echocardiography with possible secondarily involved liver.

Key Words: Blood in Vomiting, Congenital Heart Disease, Trilogy of Fallot, Congestive Heart failure

INTRODUCTION

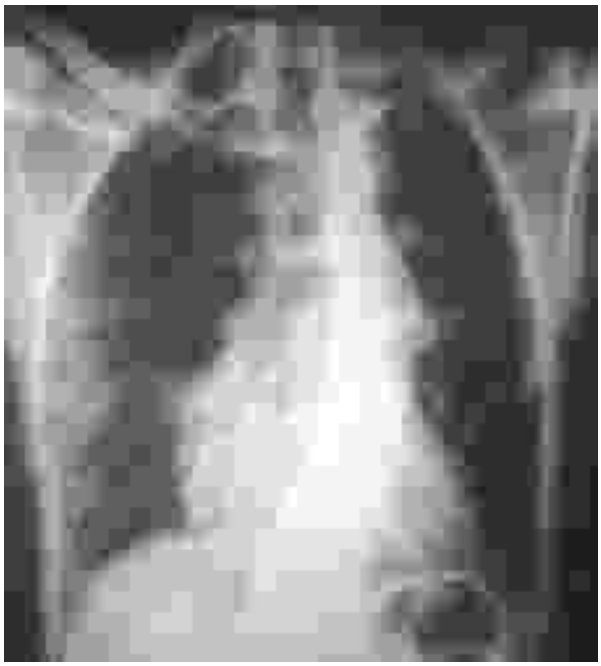
Trilogy of Fallot is a rare congenital heart condition comprising of pulmonary valve stenosis, right ventricular hypertrophy and atrial septal defect. In the paper published 1950, the combination of pulmonary stenosis with reversed right-to-left interatrial shunt without ventricular septal defect was called as "the triologie de Fallot"[1,2].

This case report highlights the importance of recognizing atypical presentation of the major problem. While considering the diagnosis of the major problems like in our case, the typical presentation are promptly recognized, but if presented with atypical features, they are often diagnosed late or underdiagnosed because physicians are often unaware of such type of scenarios. Since cyanotic heart diseases commonly present with findings related to cardiovascular system, but it should be kept in mind that they may present atypically with findings related to GI system like in our case, often if not recognized in the early stages. If physicians think in that way and perform thorough history, examination and targeted investigations, the major problems can be recognized at an earlier stages and if early intervention is done in such patients produces significant improvement in their morbidity and mortality.

CASE REPORT

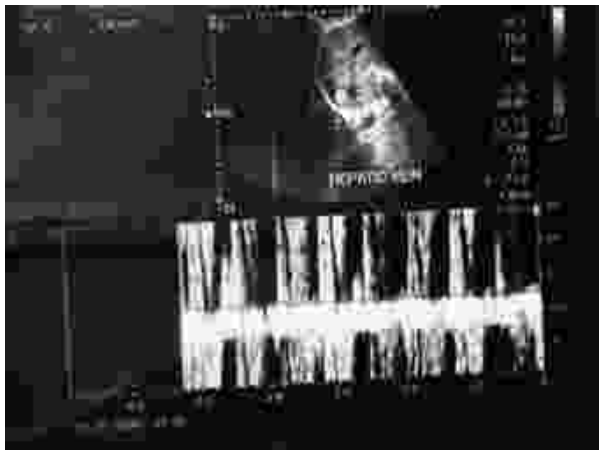
A 25 year old male, with no known co morbidities shopkeeper by profession,

Figure 1: Chest X Ray showing Kyphoscoliosis, Cardiomegaly, Limits Dilated Left Pulmonary Trunk



presented to emergency department with complaints of 2 episodes of hematamesis in the last 1 day. Vomiting was not associated with any other significant symptoms. Physical examination revealed patient to be deaf, dumb, short stature, lean built and well oriented in time, place and person. Vitals were pulse 98 bpm, BP=110/75 mmHg, Respiratory rate=16/min and afebrile. Clubbing (drum stick appearance), lip cyanosis, pectus excavatum and scoliosis were noted. Lungs were clear and a pansystolic murmur at left 2nd Intercostal space was noted. Haematological parameters which were abnormal included Hb (19.3 g/dl), HCT (61.9%) MCV (67.7 f/l), MCH (21.1 pg/cell), PLT (1,41,000/uL), INR (2.34), APTT (34.8 sec), total bilirubin (2.48 mg/dl), PH (7.42), PCO2 (27 mmhg), PO2 (50 mmhg), HCO3 (17 mEq/L), ABE (-5), LDH (339 IU/L). His

Figure 2: Doppler Ultrasound Showing to be Within Normal



TORCH profile was normal.

Endoscopy was done for suspected oesophageal varices but came to be normal. Ultrasound abdomen with Doppler was done for hepatic veins (Figure 2), CT and MRI abdomen was done to rule out any hepatic pathology but they came to be within normal limits. Chest X ray showed kyphoscoliosis, cardiomegaly, dilated left pulmonary trunk and relative hyperaemia over both lung fields (Figure 1). The 12 lead Electrocardiogram (ECG) showed normal sinus rhythm with extreme right axis deviation, tall peaked p waves in lead II, III suggesting right atrial hypertrophy, tall R waves and inverted T waves in V2, V3 and tall S wave and upright T waves in V4, V5 suggesting right ventricular hypertrophy (Figure 3). Transthoracic Echocardiography showed thickened pulmonic valve, severe infundibular pulmonic stenosis with pressure gradient of 110 mmhg, right ventricular hypertrophy, Tricuspid regurgitation (moderate to severe), Pulmonary stenosis (severe), Aortic regurgitation (mild to moderate) with normal ejection fraction. Trans oesophageal Echocardiography with bubble study revealed large atrial septal defect with right to left shunt and dilated right atrium and ventricle. Final diagnosis of Trilogy of Fallot was made

Figure 3: ECG Showing a Normal Sinus Rhythm with Peaked p Waves in Lead II, III Suggesting Right Atrial Hypertrophy, Tall R Waves and Inverted T Waves in V2, V3 and Tall S Wave and Upright T Waves in V4, V5 Suggesting Right Ventricular Hypertrophy

with possible secondary involvement of liver. He was referred to cardio surgery department for the pulmonary valvotomy and repair of ASD and recovered well. No specific events were noted at the outpatient department visits after about sufficient period of follow up.

DISCUSSION

Congenital heart diseases (CHD) can be defined as the group of disorders characterized by the defect in the structure and function of the heart. Congenital heart disease (CHD) mainly includes major structural malformations of the heart and/or major vessels which are present at, or remain persisting abnormally after birth. The prevalence of congenital heart disease (CHD) at birth has been relatively variable at 4.05 to 10.4 cases per 1000 live births in different surveys using a variety of case discovery methods. Congenital heart diseases are broadly classified into non cyanotic and cyanotic heart diseases. Cyanotic congenital heart diseases are characterized by the systemic arterial desaturation due to right to left shunt causing central cyanosis.

Congenital pulmonary stenosis commonly occur in association with the ventricular septal defect, overriding of aorta and right ventricular enlargement. This spectrum of disorder is collectively known as "Teratology of Fallot" but pulmonic stenosis, less frequently like in our case can also occur in association with intact ventricular septum, no overriding of aorta with an interatrial right to left shunt usually an atrial septal defect or patent foramen ovale. This pulmonary stenosis with a reversed interatrial shunt has been called as "triologue de Fallot" (Trilogy of Fallot). Right ventricular outflow obstruction usually resides in the stenotic mobile dome shaped pulmonary valve and occasionally due to obstruction of pulmonary arteries or its branches. The interatrial communication is usually by a patent foramen ovale or by an ostium secundum atrial septal defect. Prolonged congestive heart failure can lead to liver damage, mainly because of decreased hepatic blood flow, increased venous pressure, and decreased arterial oxygen saturation. Long-term existence of passive liver congestion, hepatic fibrosis or even cirrhosis may be developed.

Trilogy of Fallot, a very rare form of spectrum of cyanotic congenital heart disease is reported very less often in literature. Here in our case of a young gentleman who initially presented with symptoms mimicking chronic liver disease. He had an unusual appearance, physique and was dumb and deaf by birth. On thorough diagnostic work up for the suspected GI pathology, all investigations came out to be unremarkable. Further examination of patient revealed significant findings related to cardiovascular origin i.e. central cyanosis and characteristic murmurs which diverted our attention towards the possibility of cyanotic heart disease. He was investigated further, and at last, his Trans oesophageal ECHO with bubble study confirmed the diagnosis of Trilogy of Fallot. Although initial presentation was not related to final diagnosis, so there may be possibility of secondary involvement of liver due to prolonged congestive heart failure secondary to Trilogy of Fallot. As we know that cyanotic congenital heart disease if not treated at an earlier stages, can starts to cause irreversible damage to the heart and ultimately cause passive congestion, decreased blood flow and fibrosis of the liver which later can develop into cardiac cirrhosis. But his hepatic workup was absolutely normal, thus diagnostic uncertainty remains. Our case is unique in the sense that it is the first case to be reported which highlights this unusual presentation of Trilogy of Fallot. The goal behind this case report is to divert the physician's attention towards the importance of picking the unusual presentations of the major pathologies at an early stages as this would be helpful in context for benefit of patients.

CONCLUSION

The primary aim of this case report is to draw physician's attention towards this important issue of recognizing the atypical presentation of a major problem. While diagnosing any pathology like that in our case, it is necessary to perform a thorough history and examination in order not to under diagnose any major problem which may be hidden at that time unrelated to patients symptoms and may present at a later date.

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