

Pierre Robin sequence with tetralogy of Fallot: An unusual finding

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ABSTRACT

Pierre Robin sequence has been known to present with multiple somatic abnormalities. It is known that the syndrome impairs cardiogenesis. However, the reason for this is unknown. Impaired cardiogenesis manifests in the form of various defects such as ventricular septal defect and atrial septal defect but the presence of tetralogy of Fallot is very rare.

Keywords: Pierre Robin, Tetralogy of Fallot, valvular disease

Introduction

Pierre Robin sequence has set of characteristic features such as micrognathia, glossoptosis, and upper airway obstruction.^[1] It has associated cardiac defects (20%)^[2] of which ventricular septal defect (VSD) is most common. Tetralogy of Fallot (TOF) is a very uncommon presentation in this disorder and only a few studies have reported it.^[3] We discuss the case of a 17-year-old patient who is presented with signs of Pierre Robin sequence and TOF.

Case Report

A 17-year-old male presented to the outpatient department of cardiology at Rawalpindi Institute of Cardiology, Rawalpindi, Pakistan, after being referred from a local practitioner in Kashmir, for evaluation of a murmur that had been auscultated at the lower left sternal border. The patient presents with complaints of bluish discoloration of the body and difficulty in breathing for 5 years of age. The patient had a history of inconsistent follow-ups. The patient's cyanosis was exacerbated during eating and lessened spontaneously. The patient's dyspnea occurred mostly at night. Exertion was an exacerbating factor but the patient was reported that he altered his lifestyle (not taking active part in sports, taking rest after walking long distances, and walking less vigorously) accordingly to prevent exacerbation. History was not significant for any other complaints. The patient is a student of 10th grade and doing well in school. Family history is significant for consanguineous marriage of his parents. All his siblings were normal. On examination, a tall and lean male was sitting comfortably in the given settings. Anthropometry

revealed his height to be 183 cm and weight to be 40 kg. Vitals were all normal. There was bilateral Grade 3 clubbing on both his fingers and toes [Figure 1].

There was central cyanosis on the lips. Micrognathia was also noted [Figure 2].

On precordial examination, the lateral diameter of the chest was lesser and not in proportion to anteroposterior diameter. Apex beat was found in fifth intercostal space in the mid-clavicular line. It was normal in character. Auscultation revealed normal heart sounds and a holosystolic murmur, Grade 3/6, heard at the lower left sternal border. Rest of history and examination was unremarkable.

Pediatrics department was also consulted. A provisional diagnosis of Pierre Robin sequence with VSD was made. The patient was prescribed oral bosentan, 62.5 mg once daily, sildenafil, 5 mg thrice daily, and spironolactone, 40 mg once daily. Echocardiography could not be attempted due to rib crowding. However, his computed tomography (CT) images revealed characteristic findings of TOF [Figures 3 and 4].

VSD correction with infundibular repair was attempted and the patient was subsequently discharged. He had no post-operative complications.

Discussion

This is the first case report from Pakistan discussing TOF in a patient with previously undiagnosed Pierre Robin sequence. The



Figure 1: Clubbing noted on both fingers. Schamroth's sign elicited



Figure 2: Micrognathia

patient did not have a cleft palate^[4] which completes the classic triad but the syndrome was diagnosed considering the patient's presenting symptoms and features. A recent study shows that mutation in *BMPR1B* gene^[5] is not only responsible for Pierre Robin sequence but also pulmonary hypertension.^[6] However, it does not explain the pulmonic stenosis found in our patient.

The case report emphasizes the need for evaluating all patients with appropriate radiological investigations for clinical presentations might be misleading as was the case for our patient. Clinical findings were significant for a murmur of VSD while radiological evidence showed TOF. The need for thorough radiologic investigation^[7] has been emphasized by other studies as well. TOF is usually diagnosed on chest X-ray with its classical findings but echocardiogram continues to be both the diagnostic and therapeutic investigation.^[8] In our report, CT scan was performed because the cardiac defect that had been anticipated was a VSD and not TOF.

There are multiple genes that have been implicated in development of TOF.^[9] These genes are also a part of the spliceosome family^[10] responsible for Pierre Robin sequence and other syndromes in which severe rib crowding occurs. This

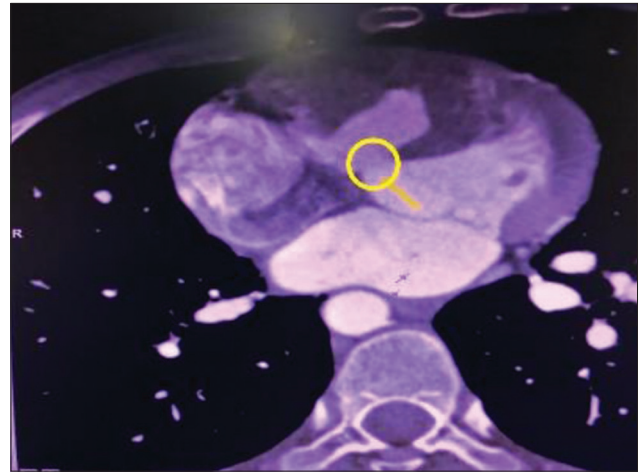


Figure 3: AP view in computed tomography scan showing ventricular septal defect (yellow circle)

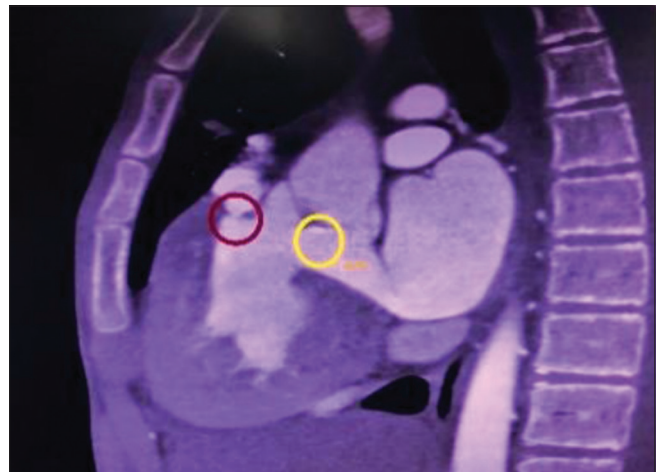


Figure 4: Lateral view in computed tomography showing pulmonic stenosis (red circle) and overriding aorta (yellow circle)

genetic theory provides a possible explanation as to how severe rib crowding coexists in a patient with Pierre Robin sequence.

It is also important to note that therapeutic interventions are not based on correcting the hemodynamic abnormality as a whole but on the most symptomatic lesion, as in the case. There are multiple methods to correct the defect but infundibular repair^[11] is the mainstay of all these procedures. The outcome in most cases is positive except in cases of severe right-sided flow obstruction. In these exceptions, patients have to be reoperated but our patient did not have such severe issues and, therefore, had no complications.

Conclusion

In patients who present with features of a somatic disorder and a murmur, TOF should be considered as an important cardiac defect after accounting for VSD. It must also be remembered that TOF may not present as a classic ejection systolic murmur and might present as holosystolic murmur in these cases. The patients with such murmurs need urgent radiological

investigation for timely intervention before complications, such as pulmonary hypertension, develop.

Authors' Declaration Statements

The authors' guarantee that the work is original and does not infringe copyright or other party's property rights. All authors have read and approved this submission and have given appropriate credit to everyone who participated in this work.

Ethics Approval and Consent to Participate

Patient consent

The patient's consent was taken before reporting the case.

Availability of data and material

The data used in this study are available and will be provided by the corresponding author on a reasonable request.

Competing interest

None to declare.

Funding statement

None to declare.

Authors' contributions

Nismat Javed wrote the first draft, critically revised and wrote the final version of manuscript. Jahanzeb Malik created the idea, provided case information and also revised the final draft. Both authors approve the final version of the study.

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