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Late Presentation of Partial Atrioventricular Septal Defect: A Case Report

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Abstract

Atrioventricular septal defect is a spectrum of congenital heart defects that can be classified as either complete or partial with the complete form being the most common and usually present in the neonatal period with congestive heart failure. However, partial defects can remain asymptomatic and present later in adulthood with variable degrees of heart failure. The case under investigation is a middle aged female with a 2-year history of worsening dyspnea and recurrent chest infections due to undiagnosed partial atrioventricular septal defects. As a conclusion, it is important to establish a screening program to detect the asymptomatic cases early and prevent delay consequences.

Key words: partial atrioventricular septal defect, adult, screening

Background

Atrioventricular septal defect (AVSD) is a spectrum of congenital heart diseases that can have varying degrees of severity ranging from a clinical presentation of congestive heart failure in the neonatal period to a subtle disease that can present later in life and with less severe symptoms, depending on the size and nature of defect [1]. It results from a congenital defect in the endocardial cushion which is responsible for atrial and ventricular septation and formation of atrioventricular (AV) valves [2]. Such a defect can be complete, i.e., ostium primum atrial septal defect (ASD), common AV valve and inlet ventricular septal defect, or partial [3]. Partial AVSD (pAVSD) can be defined as ostium primum ASD and a cleft in the anterior mitral leaflet[4]. AVSD constitutes around 5-8% of the estimated cardiovascular malformations with the complete form most commonly linked to major chromosomal abnormalities and Down syndrome being on top. On the other hand, pAVSD is more common in patients without Down syndrome [5,6].

This report presents a case of 52 years old female who came to the outpatient clinic complaining, for 2 years duration, of shortness of breath on exertion. After clinical examination and routine investigation including a transthoracic echocardiography (TTE), she was diagnosed with large ASD primum, clefted anterior mitral valve leaflet with mitral and tricuspid regurgitation. Surgery was done to repair the defect and post-operative period was uneventful.

The decision to share this case was to discuss the importance of establishing a

screening method for congenital heart diseases and to draw attention to the fact that congenital heart diseases should be kept in mind as part of the differential diagnosis of dyspnea even in older age groups.

Case presentation

A 52-year old diabetic and hypertensive housewife, height: 145 cm, weight: 61 kg, presented to an outpatient clinic with a history of shortness of breath on minimal activity (NYHA class III) associated with orthopnea. Two years before her presentation, her condition started as chest infection that led to a slow progressive feeling of tiredness and shortness of breath with activity. The patient was completely well and fit before this episode with very good exercise tolerance. She reported worsening of symptoms with time and associated recurrent chest infections, palpitation and chest pain. She sought medical attention and general and cardiorespiratory examinations that showed pansystolic murmur at mitral and tricuspid areas and coarse crackles at both lung bases. On further questioning, the patient admitted having abdominal discomfort; examination showed grade 1-2 pitting lower extremity edema. Electrocardiography (ECG) showed a PR interval prolongation (240 ms) and left axis deviation, findings consistent with her later diagnosis of pAVSD [7] (Fig. 1). Chest x- ray showed an increased cardiothoracic ratio with pulmonary congestion (Fig. 2A). TTE showed a large ASD primum with dilation of the left and right atria, right ventricle, and normal ejection fraction with mitral and tricuspid regurgitation (Fig. 3). Hence, the patient was referred for coronary and cardiac chamber catheterization. While coronary angiography was completely normal, cardiac chamber catheterization showed a pulmonary artery systolic pressure of 10 mmHg. Open cardiac surgery was accordingly planned, see details in Intraoperative notes; Figure (2B) shows postoperative chest x- ray.

As a follow up for the patient, an appointment was done after one month of surgery and the patient was recovering very well; TTE showed no residual mitral regurgitation, closed ASD with no leak and good left ventricular function. After 3 months, the patient was in good health with no reported complains.

Preoperative and surgical interventions

In the preoperative period, all necessary biochemical and serological investigations were done; findings are shown in Table (1). ECG and Echocardiography were repeated and showed the same results as before.

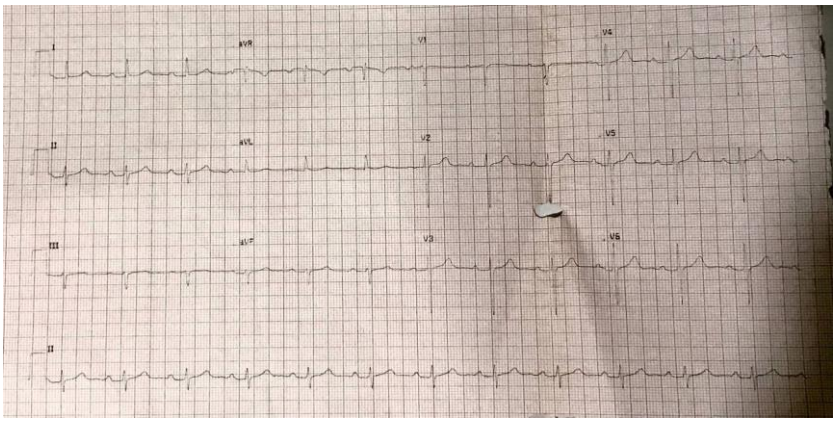

Figure 1: ECG showing prolonged PR interval (first degree AV block) and left axis deviation

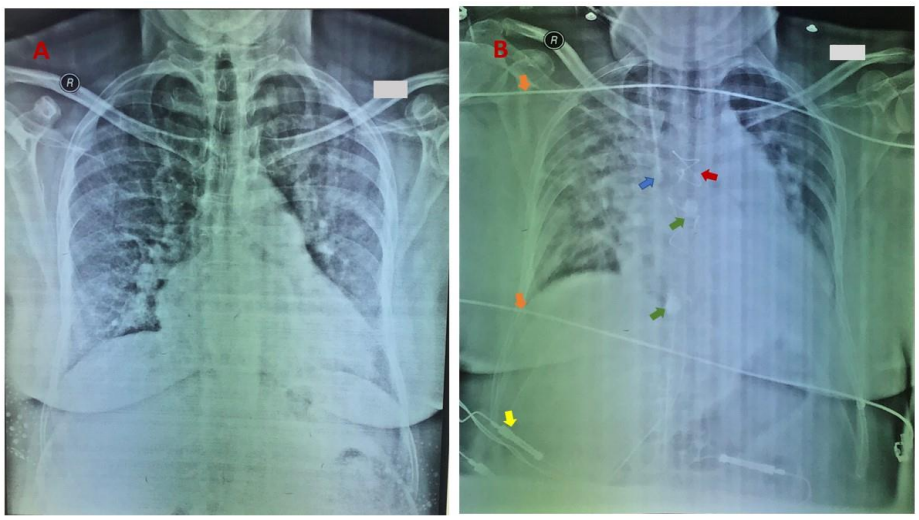

Figure 2: chest x- ray posteroanterior view showing pulmonary congestion and increased cardiothoracic ratio (both A and B); preoperative x- ray (A); post- operative x- ray showing steel- wire for closure of median sternotomy (red arrow), chest drains (green arrows), central venous line (blue arrow), pacemaker wire (yellow arrow), and ECG lead wires (orange arrow) (B).

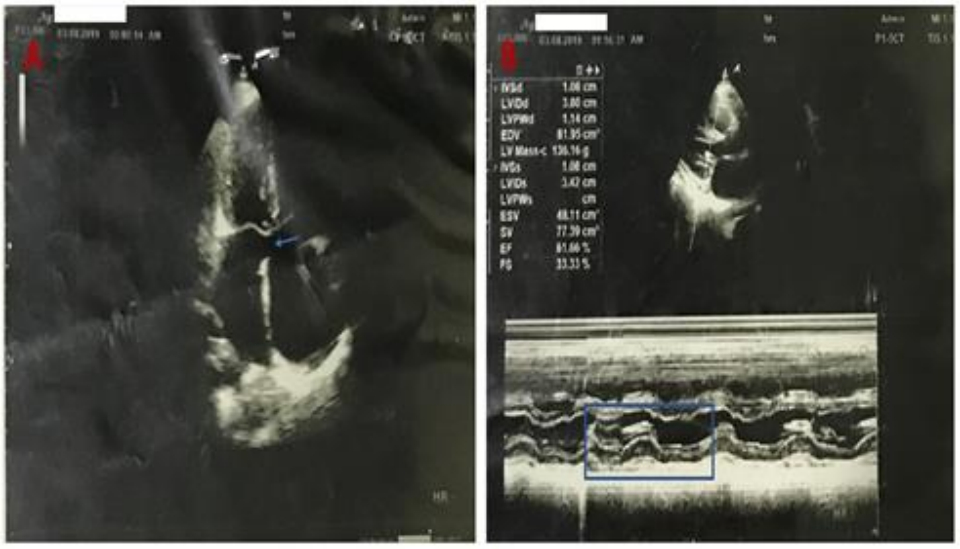

Figure 3: Apical four chamber view showing large ASD primum (line arrow). (A) parasternal long axis view with M-mode showing abnormal movement of mitral valve leaflets and inadequate coaptation (selected area in B).

Table 1: High random blood sugar and ALP; ALP, alkaline phosphatase; ALT, alanine aminotransferase; AST, aspartate aminotransferse; TSB, total serum bilirubin

Indices	Value
Random blood sugar	233 mg/dl
AST, ALT, ALP	36, 33, 372 U/L respectively
TSB	0.8 mg/dl
Blood urea and serum creatinine	32 and 0.5 mg/dl respectively
Viral screen	Negative

Preoperative and surgical interventions

In the preoperative period, all necessary biochemical and serological investigations were done; findings are shown in Table (1). ECG and Echocardiography were repeated and showed the same results as before.

Intraoperative notes

A surgery was done under general anesthesia with ECG and blood pressure monitoring all on set. The chest wall was opened in layers via median sternotomy then the pericardium was opened through longitudinal pericardiotomy, followed by classical cannulation with single aortic cannula and two separate venous cannulae in the superior and inferior vena cava (SVC and IVC) after administration of intravenous heparin. Cardioplegia cannula was inserted in the aortic root. A total cardiopulmonary bypass was initiated by cross clamping the aorta and arresting the heart via infusion of cardioplegia (Del Nido). The patient was cooled down to 30°C and the heart was immersed in iced water.

The heart was accessed via right atriotomy; then, venting the pulmonary venous return was done with a small vent. An assessment and evaluation of morphology of the defect was done and a primum ASD with cleft in the mitral valve and regurgitation in both AV valves was noticed. The coronary sinus was found intact with no evidence of left superior vena cava (LSVC). The cleft in the mitral valve was repaired by directly interrupted simple suturing using proline 5 zero. The ASD was closed completely by using a non- tanned autologous pericardial patch which was sutured using proline 4 zero. The tricuspid valve was repaired via the suture bicuspidization of the inferior and septal cusps with proline 5 zero. An atrium was closed in two layers and the aortic

cross clamp was removed after careful de-airing. The heart started beating spontaneously; the cardiorespiratory bypass was stopped, and the heparin effect was reversed with protamine sulfate. By then, de-cannulation and thorough hemostasis were achieved and two mediastinal drains were inserted before closing the sternum with steel wires and the rest of the chest wall in layers. The patient was then transferred to the intensive care unit (ICU) for following up and close-monitoring.

In the ICU, the progression was uneventful and the extubation was done on the same day. The cardiac support started in the theatre and continued throughout the ICU period with gradual declining till total weaning on the third postoperative day. Postoperative echocardiography is shown in Figure (4).

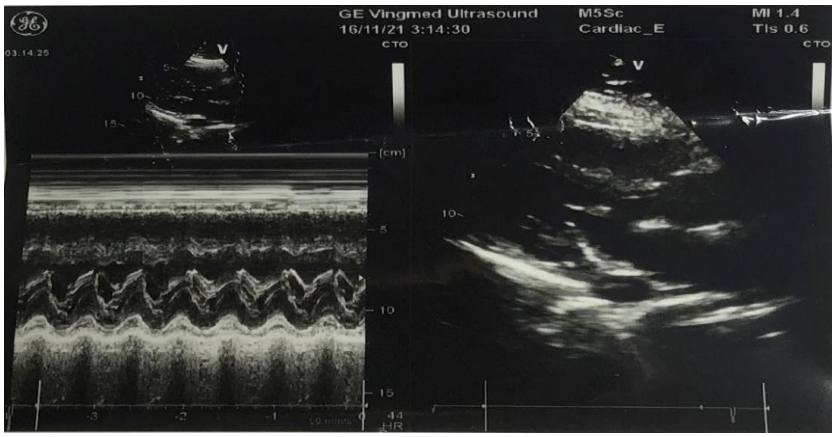

Figure 4: Postoperative echocardiography with parasternal long axis view and M-mode *showing normal movement of mitral valve leaflets and proper coaptation.*

Discussion

This case, like many others which represent a late and undergo surgical correction with immediate good response and marked improvement of patients' conditions in the early postoperative period, gave rise to some loud questions: what happens later? Is this a natural history to go undetected until adulthood? Can surgery fix the problem at any time?

Review of literature related to repair of pAVSD in pediatric patients shows no significant difference in outcome by child age, regarding postoperative mortality and the need for reoperation. The major concern in very young children (1-2 years) is the need for blood transfusion during surgery in those under 15 kg, in addition to the postoperative feeding difficulty that can prolong postoperative hospitalization. Meanwhile, in elder children, the drawback of delay is the risk of developing an AV annular dilation along with left atrial and ventricular dilation that can make repair more difficult. So, from a benefit risk perspective the recommendation was to repair pAVSD between 1-2 years of age [8].

A lot of work has been done to study repair of pAVSD in adults and mostly agree on good outcomes of surgery with no significant surgical mortality as in the present case. However, many problems could accompany late diagnosis and repair, including arrythmia due to heart chamber dilation and remodeling, and increasing the need for left AV valve annuloplasty and replacement with associated increased risk of complete heart block. The need for reoperation in adults is a big concern with 76% and 21% of patients needed reoperation for left AV valve regurgitation and left ventricular outflow tract obstruction, respectively, between 5-10 years postoperatively compared to only 5% and 6% for the same aforementioned reasons in the pediatric study [7,8.9]. So early repair in childhood is always the preferred option.

Out of the data above, one can see a great necessity to find a method to identify these individual cases early in life and prevent delay consequences. Regarding screening for congenital heart disease, a screening program with pulse oximetry for detection of hypoxemia before neonatal discharge is now considered routine for detecting critical congenital heart diseases [10]. Furthermore, prenatal and early postnatal echocardiographic screening is used in groups with a high risk of developing congenital heart disease like those with Down syndrome and other high-risk syndromes and those with abnormal in utero ultrasound findings[11]. Some data discuss the utility of echocardiographic screening for the general population considering it as part of clinical neonatal examination before discharge. However, a high false positive was seen in this very early age which may lead to unnecessary intervention in addition to problems regarding the cost of applying such a screening program on a wide base [12,13]. Further, a study needs to be done to assess the significance of echocardiographic screening after 6 months of age to detect asymptomatic lesions that need early repair.

Conclusion

The undiagnosed congenital heart disease is an important differential diagnosis to keep in mind in adults who present with symptoms of heart failure with no current signs or previous history of ischemic heart disease. More work is needed to establish a well-organized screening program to diagnose these cases early in life and to prevent complications associated with delayed diagnosis.

Conflict of interest

We declare that one of the authors, Dr. Wadhah Mahbuba, is a member of the editorial board in Kufa Medical Journal.

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