Partial Atrioventricular Septal Defect in a Case of Bardet-Biedl Syndrome: A Rare Association

Mohaddeseh Behjati[1], Fatemeh Tohidi[2,3], Mohammad Karimian[4]

[1] *Rajaie Cardiovascular Medical and Research Center, Iran University of Medical Sciences, Tehran, Iran*

[2] *Cancer Research Center, Health Research Institute, Babol University of Medical Sciences, Babol, Iran*

[3] *Cellular and Molecular Biology Research Center, Health Research Institute, Babol University of Medical Sciences, Babol, Iran*

[4] *Department of Molecular and Cell Biology, Faculty of Basic Sciences, University of Mazandaran, Babolsar, Iran*

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Abstract- Bardet-Biedl syndrome (BBS) (MIM 209900) is a genetic disorder with a wide spectrum of clinical manifestations including retinal dystrophy, hypogenitalism, polydactyly, obesity, renal abnormalities and mental retardation. We describe a 13-year-old girl, a known case of Bardet-Biedl syndrome, who was going to undergo hysterectomy due to hydrometrocolpous. She was homozygous autosomal recessive for gene BS57. She was obese and had impaired vision, renal abnormality, borderline intelligence, recurrent urinary tract infection, menstrual problems, normal secondary sex chrematistics and corrected polydactylies. She had also big nose, thin upper lip, slightly everted lower lip, small mouth and retrognathia. Her electrocardiography showed incomplete right bundle branch block. We identified atrioventricular septal defect (AVSD). In conclusion, physicians who deal with cases who suffered from Bardet-Biedl syndrome, should be vigilant about seeking for identification of cardiac anomalies such as partial AVSD. This leads to earlier identification of the existing cardiovascular disease which facilitates appliance of curative measures.

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Keywords: Bardet-biedl syndrome; Partial atrioventricular septal defect; Congenital heart disease

Introduction

Bardet-Biedl syndrome (BBS) (MIM 209900) is a genetic disorder with a wide spectrum of clinical presentations including obesity, retinal dystrophy, hypogenitalism, polydactyly, renal abnormalities and mental retardation (1). Hereby, we present a known case of BBS with partial atrioventricular septal defect (AVSD), a previously undescribed feature in this setting.

Case Report

Our case was a 13-year-old girl who referred us for pre-operation cardiac consult. She was going to undergo hysterectomy due to hydrometrocolpous. She was a known case of Bardet-Biedl syndrome (#615984) which was genetically confirmed through karyotyping. Her genetic test showed homozygous autosomal recessive for the gene BS57 which was pathogenic. Her parents had consanguineal marriage. She was obese and had impaired vision, renal abnormality, borderline intelligence, polydactylies which was surgically corrected at her childhood, recurrent urinary tract infection and menstrual problems but with normal secondary sex chrematistics. She had some reported facial features of BBS cases such as big nose, thin upper lip, slightly everted lower lip, small mouth and retrognathia (Figure 1). The referring physician identified an atrial septal defect by performing trans thoracic echocardiography (TTE). Her electrocardiography showed incomplete right bundle branch block (Figure 2). We identified atrioventricular septal defect (AVSD) using TTE and confirmed it by trans esophageal echocardiography (TEE). There was also mild to moderate eccentric mitral regurgitation due to anterior mitral leaflet prolapse. TEE views are depicted in Figure 2.

Corresponding Author: M. Behjati
Rajaie Cardiovascular Medical and Research Center, Iran University of Medical Sciences, Tehran, Iran
Tel: +98 2142910700, E-mail address: dr.mohaddesehbehjati@gmail.com

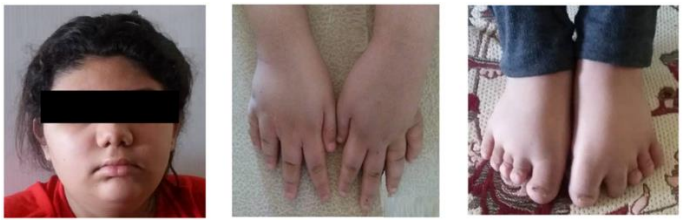

Figure 1. Facial properties of BBS subject. A big nose, thin upper lip, slightly everted lower lip, small mouth and retrognathia

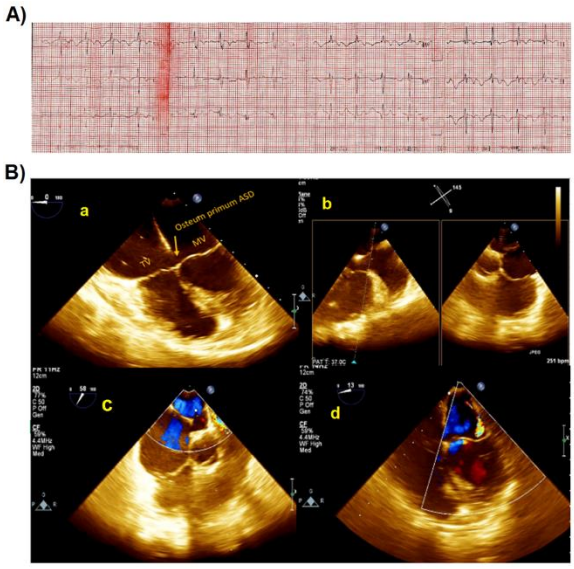

Figure 2. Electrocardiography strip of the case and the electrocardiogram of the patient shows incomplete right bundle branch block. A) Transesophageal echocardiography views of partial atrioventricular septal defect. B) Transesophageal four chamber view which shows primum atrial septal defect (a). X-plain transesophageal view of interatrial septum which shows absence of any other atrial septal defects (b). Transesophageal view at 600 which shows primum atrial septal defect (c). Transesophageal four chamber view which shows mild to moderate eccentric mitral regurgitation due to prolaptic anterior mitral valve without any cleft (d)

Discussion

Partial AVSD is characterized by the presence of ostium primum atrial septal defect with intact ventricular septum (2). This might result in right ventricular overload (3). The referral for surgical correction of affected children with partial AVSD is often delayed due to their being largely asymptomatic (3). This congenital defect could occur both in isolation and with other congenital diseases (4). By now, various associations rather than cardinal features of BBS have been reported such as Hirschsprung disease, multiple pigmented naevi, situs inversus, epilepsy, hearing impairment and diabetes mellitus (5,6). To best of our knowledge, the association of partial AVSD and BBS is very rare. In conclusion, physicians who deal with cases who suffered from BBS, should have especial considerations to cardiac anomalies such as partial AVSD. This leads to earlier identification of the existing cardiovascular diseases which facilitates appliance of curative measures at earlier stages.

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