

Repair of double orifice mitral valve with an atrioventricular septal defect in a girl with Ellis-Van Creveld syndrome

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Abstract

Ellis-van Creveld syndrome is a rare autosomal recessive disorder. We describe the case of a 7-year-old girl with Ellis-van Creveld Syndrome with the diagnosis of common atrium and partial atrioventricular septal defect. She underwent a successful surgical repair, and intraoperatively, a double orifice mitral valve was diagnosed as well.

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Abstract:

Ellis-van Creveld syndrome is a rare autosomal recessive disorder caused by mutations in the EVC and EVC2 genes. The four principal manifestations are chondrodysplasia, polydactyly, ectodermal dysplasia and congenital heart defects. We describe the case of a 7-year-old girl with Ellis-van Creveld Syndrome with the diagnosis of common atrium and partial atrioventricular septal defect. She underwent a successful surgical repair, and intraoperatively, a double orifice mitral valve was diagnosed as well. The correct diagnosis of this disorder in early life is essential in the overall prognosis of the syndrome. Clinical follow-up at regular intervals is very important in these patients to institute proper managements and prevent further complications.

Key words:

Ellis-Van Creveld syndrome, common atrium, partial atrioventricular septal defect, double-orifice mitral valve.

Key Clinical Message:

Congenital heart defects are common among Ellis-Van Creveld syndrome patients, and need to be managed to enhance the quality of life and improve prognosis. Partial atrioventricular septal defect is among the most common congenital heart defects in these patients, and double orifice mitral valve is encountered frequently.

Introduction:

Ellis–van Creveld (EVC) syndrome is a rare skeletal and ectodermal dysplasia, with a prevalence of about 1 in 20,000 births, and is common among the Old Order Amish of Lancaster County, Pennsylvania (1). The skeletal dysplasia presents at birth with short limbs, and affected individuals have postaxial polydactyly of the hands and sometimes of the feet, and dysplastic teeth and nails (1). This condition was first described by Richard Ellis and Simon Van Creveld in 1940 (2). Congenital heart defects (CHD) are encountered in about 50-60 % of the cases, and include common atrium, atrioventricular septal defect (AVSD) with anomalies of the mitral and tricuspid valves, patent ductus arteriosus, ventricular septal defect, atrial septal defect, and hypoplastic left heart syndrome (3). Moreover, systemic and/or pulmonary venous abnormalities are common as well (4). Double orifice mitral valve (DOMV) is a rare congenital anomaly of the subvalvar mitral valve apparatus in which there is an accessory bridge of fibrous tissue that partially or completely divides the mitral valve into two orifices which functionally may result in regurgitation or stenosis of the mitral valve. It has been reported that AVSD was the most common CHD associated with DOMV (5). Herein, we introduce the case of a 7-year-old girl with EVC Syndrome who underwent open heart surgery for repair of partial atrioventricular septal defect (PAVSD) with DOMV and common atrium.

Case Report:

A 7-year-old girl presented to our children hospital with exertional dyspnea and easy fatigability and frequent respiratory tract infections. The patients did not have any significant antenatal, natal, and neonatal history. She had normal intelligence and was doing well at school. The lips and fingernails were mildly cyanotic without clubbing. The oxygen saturation was 85% by pulse oximetry on room air. Arterial blood gas on room air showed hypoxia with PaO₂ of 50 mmHg and SaO₂of 83%. On general examination, she had short stature, narrow thorax, short fingers and toes, bilateral polydactyly of hands (Figure 1) and left foot. Fingernails and toenails were markedly hypoplastic, thin and wrinkled. In oral examination, anterior teeth were conical. Examination of the cardiovascular system revealed a 3/6 short systolic murmur at the left lower sternal border and a loud second heart sound. The electrocardiogram revealed left axis deviation, with complete right bundle branch block in leads V1,V2. The chest X-ray showed cardiomegaly with dilated main pulmonary artery and its branches. Transthoracic echocardiogram (TTE) showed common atrium and PAVSD with moderate mitral regurgitation (MR), mild tricuspid valve regurgitation, and moderate pulmonary hypertension. A clinical diagnosis of EVC syndrome was established. Based on the pre-operative findings, the patient was planned for surgical repair of PAVSD and common atrium. The operation was performed through median sternotomy. Total cardiopulmonary bypass was prepared, the aorta was cross-clamped, and the heart was arrested by antegrade cardioplegic solution. A right atrial incision was made parallel to the right atrioventricular groove, and the intracardiac anatomy was explored. There was DOMV (Figure 2), which was not diagnosed by preoperative TTE. The greater orifice of the mitral valve was similar to that in the classic PAVSD, and consisted of three leaflets (left superior leaflet (LSL), left inferior leaflet (LIL), and left lateral leaflet). This orifice (The greater one) of the mitral valve was repaired by suturing the free edges of LSL and LIL to each other by fine 6/0 Prolene sutures, thus converting it to a bileaflet valve (Figure 3). The other small orifice was left intact to avoid any possible mitral stenosis. Saline test showed excellent result with trivial residual MR. There was complete absence of the atrial septum without any remnants. A new atrial septum was constructed by an autologous fresh pericardial patch. The remainder of the operation progressed uneventfully and without any conduction disturbances. Postoperative TTE showed no residual shunt across the new atrial septum, and trivial MR. The postoperative period was uneventful, and the arterial blood gas showed normal oxygenation. The patient was discharged in stable condition, and has been on regular follow-up for last 6 months without any complains.

Discussion:

EVC syndrome is an autosomal recessive syndrome, and 30% of patients die during the first year of life due to cardiac or respiratory problems (1). A retrospective review of the cases submitted to the Pediatric Cardiac Care Consortium between 1982 and 2007 was performed, and included thirty-two pediatric patients with CHD and EVC syndrome (4), and of whom twenty-eight (88%) had an AVSD, with 15 having common

atrium, and two had DOMV (4). The medical literature reported that there are three morphological types of DOMV: central type, duplicate mitral valve type, eccentric type. The most common type is the eccentric one which is characterized by a large main opening and a smaller accessory opening (6), which was the type we found during our surgery, and was associated with PAVSD and common atrium. Regarding the ideal time for surgical repair of CHD in patients with EVC syndrome, it has been reported that delayed surgical repair was associated with reduction in postoperative morbidity and improved survival (7). In our case, the patient was referred to our hospital at the age of seven years, and the decision of surgical repair was taken by the heart team. She remains completely asymptomatic 6 months after surgery. The management of EVC required the coordinated efforts of a multidisciplinary team of medical professionals. We made a plan with our team of specialists to systematically and comprehensively treat the patient. Moreover, genetic counseling for the patient and her family is recommended.

Conclusion:

Congenital heart defects are common among EVC syndrome patients, and need to be managed to enhance the quality of life and improve prognosis. PAVSD is among the most common CHD in these patients, and DOMV is encountered frequently and needs special attention.

Figure legends:

Figure 1: Image of the patient's hands showing bilateral polydactyly.

Figure 2: Intraoperative image showing the double orifice mitral valve; the greater arrow indicates the greater orifice, the smaller arrow indicates the smaller orifice

Figure 3: Intraoperative image showing the repaired mitral valve with complete competence.

Author Contribution

Alwaleed Al-Dairy : Planned and performed the work leading to the report. Wrote and reviewed successive versions and participated in their revisions.

Samir Srour : Participated in writing the report and approved the final version

Alaa Chaker : wrote and reviewed the successive versions and participated in their revisions

Fatima Alzahraa Alsyed Hasan : wrote and reviewed the successive versions and participated in their revisions

Author's Statement:

Consent: Written informed consent was obtained from the patient's parents to publish this report in accordance with the journal's patient-consent policy.

Author's Declaration:

None of the authors listed on the manuscript are employed by a government agency that has a primary function other than research and/or education. Moreover, none of the authors are submitting this manuscript as an official representative or on behalf of the government.

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The authors have no conflict of interest

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