Singapore Med J 2007; 48(7) : 684

A rare variant of Ellis van Creveld syndrome

Chakraborty P P, Bandyopadhyay D, Mandal S K, Subhasis R C

ABSTRACT

A nine-year-old boy presented with progressively-increasing exertional dyspnoea for the last three months. The only significant finding in the general survey was polydactyly. His vital signs were normal. He had a prominent apical diastolic thrill, a prominent S1 with a low-pitched grade 4/6 mid diastolic rumbling murmur over the apex. The S2 was widely split, fixed and the second component was louder than the first one. There was a grade 3/6 ejection systolic murmur over the left second intercostal space. Electrocardiography showed features of left axis deviation, bi-atrial enlargement and right ventricular hypertrophy. Transthoracic echocardiography identified a thin undulating intra-atrial membrane on the left side along with an ostium-primum defect. In this patient, the diagnosis of a variant of Ellis van Creveld syndrome was made.

Keywords: cor triatriatum sinister, Ellis van Creveld syndrome, heart-hand syndrome, ostium-primum defect, polydactyly

Singapore Med J 2007;48(7):684–686

INTRODUCTION

Cor triatriatum (CT) is a rare congenital cardiac anomaly with a prevalence of about 0.1% of all cases of congenital heart disease. Classical Ellis van Creveld (EVC) syndrome comprises a tetrad of clinical manifestations characterised by chondrodystrophy, polydactyly, ectodermal dysplasia, and cardiac defects. However, clinical presentation is variable, and the full spectrum may be lacking in any particular patient. A male child with cor triatriatum sinister (CTS), partial endocardial cushion defect and polydactyly, is being reported. This patient likely represents an unusual variant of EVC syndrome. To the best of our knowledge, only one patient with such an association has been previously documented.[1]

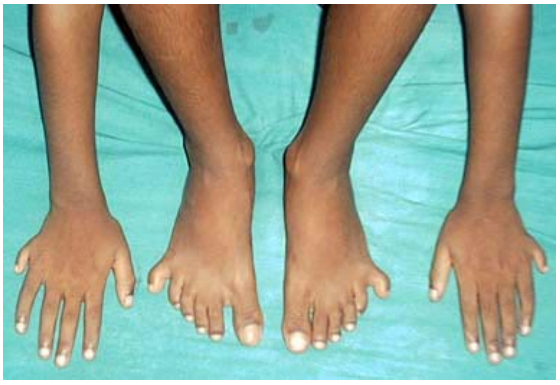

Fig. 1 Clinical photograph shows polydactyly of all four limbs.

CASE REPORT

A nine-year-old boy was brought to us with shortness of breath and heart palpitations. The boy had been suffering from frequent respiratory infections since his childhood and had developed progressively increasing exertional dyspnoea for the last three months. At presentation, the child was Class II symptomatic. There was no history of paroxysmal nocturnal dyspnoea, haemoptysis, and chest pain, swelling of legs, syncope, dizziness, squatting or bluish discolouration. His parents denied features of rheumatic fever in the past. The child was born of non-consanguineous marriage and his mother had an uncomplicated pregnancy. He had normal developmental milestones and the family history was non-contributory.

The significant finding in the general survey was mild gingival hypertrophy and polydactyly (Fig.1). His vital signs were normal with a blood pressure of 110/70 mmHg. The pulse was of normal volume and had a regular rate of 90/minute. The jugular venous pressure was not elevated. Apex beat was situated in the left fifth intercostal space just outside the mid-clavicular line, and he had a prominent apical diastolic thrill. Auscultation revealed a prominent S1 with a low-pitched grade 4/6 mid-diastolic rumbling murmur (MDM) over the apex. It was more prominent in left lateral decubitus and at the height of expiration. A grade 2/6 holosystolic apical murmur was also evident. The S2 was widely split, fixed and the second component was louder than the first one. There was

Department of Medicine, Midnapore Medical College, PO Midnapore, Dist. Pashim Medinipur, West Bengal 721101, India

Chakraborty PP, MD
RMO and Clinical Tutor

Department of Medicine, Medical College, 88 College Street, Kolkata 700073, West Bengal, India

Bandyopadhyay D, MD
Assistant Professor

Mandal SK, MD
Assistant Professor

Subhasis RC, MBBS
Postgraduate Trainee

Correspondence to:
Dr Partha Pratim Chakraborty
BE 64,
Bidhan Nagar (East),
PO Midnapore,
Dist. Paschim Medinipur
West Bengal 721101,
India
Tel: (91) 98300 92947
Email: docparthapc@yahoo.co.in

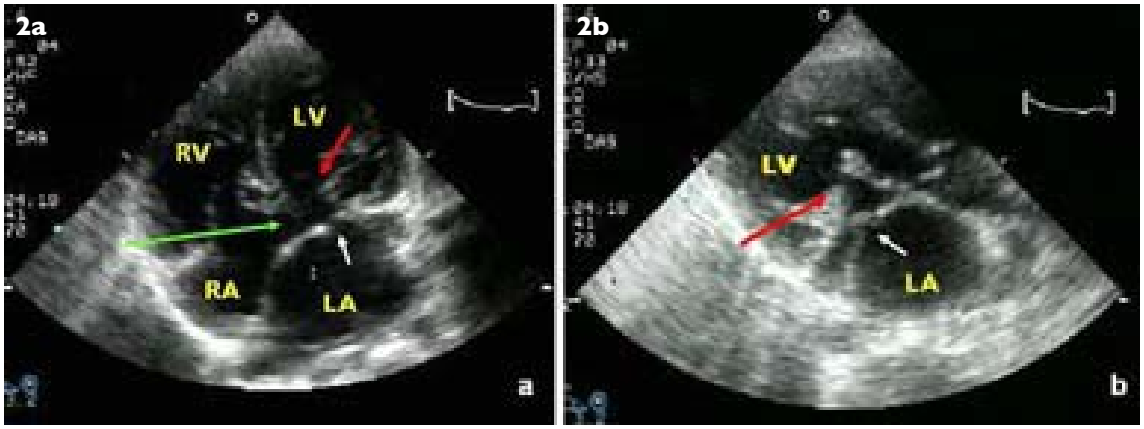

Fig. 2 Transthoracic echocardiographical (a) apical four chamber and (b) parasternal long axis images show a membrane in the left atrium (LA) with central fenestration (white arrow) and ostium-primum defect (green arrow). The mitral valve is opening normally (red arrow).

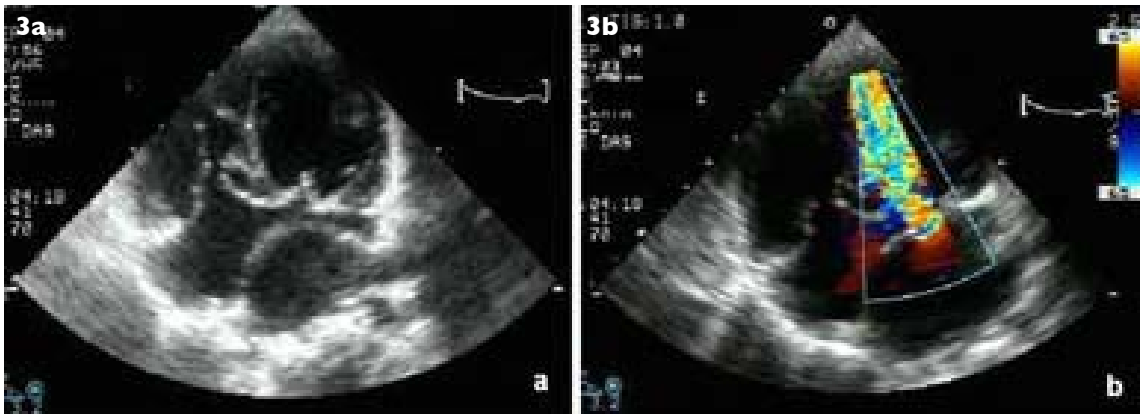

Fig. 3 Transthoracic echocardiographical (a) apical four chamber image shows the defects and (b) colour Doppler image shows turbulent flow across the fenestration of the intra-atrial membrane.

a grade 3/6 ejection systolic murmur over the left second ICS without any preceding click. Examination of the other systems did not reveal any abnormality.

The complete blood count and the baseline biochemistry were within normal limits. Chest radiograph documented a slightly increased cardiothoracic ratio with normal lung fields. Electrocardiography showed features of left axis deviation, bi-atrial enlargement and right ventricular hypertrophy. Transthoracic echocardiography identified a thin undulating intra-atrial membrane on the left side along with an ostium-primum defect (Fig. 2). It was associated with the characteristic feature of diastolic movement of the membrane towards the mitral valve and systolic movement away. Colour flow imaging and Doppler study revealed obstruction to left atrial flow (Fig. 3), left-to-right shunt across the septal defect, along with mitral and tricuspid regurgitation. The mean pulmonary artery pressure (MPA) was measured using the Doppler gradient of the tricuspid and pulmonary regurgitant jet. There was evidence of mild pulmonary hypertension (MPA 22 mm).

DISCUSSION

CTS is a rare congenital cardiac malformation consisting of a fibromuscular membrane that divides the left atrium into a postero-superior (or accessory) chamber and an antero-inferior chamber (true left atrium, containing the left atrial appendage). In its classic form, the accessory chamber receives the pulmonary veins and communicates with the left atrium by one or more fenestrations in the membrane. The malformation is usually isolated, but in about one in four patients, it is associated with other congenital defects of a complex nature.[2] CT can be classified into the following subtypes: type I is the classical form with intact atrial septum, type IIa has an atrial septal defect (ASD) between the accessory and the right atrial chambers; type IIb has ASD between the right and the true left atrial chambers,

and type IIc is a complex form that has both communications of types IIa and IIb.[3] Our patient had type IIb CT (Fig. 2).

Atrioventricular septal defect with CT is a rare combination with very few documented cases.[4] In the recent past, three cases of CT with partial AV canal defect have been reported from India.[5] Heart-hand syndromes, a genetically-heterogeneous family of disorders, comprise a class of combined congenital cardiac and limb deformities, and the prototypical heart-hand disorder is Holt-Oram syndrome.[6,7] Our patient had polydactyly with congenital cardiac defects which may be an unusual variant of the so-called heart-hand syndrome like the EVC syndrome or its variant.

EVC syndrome is a rare autosomal recessive disorder. Classical EVC syndrome comprises a tetrad of clinical manifestations of chondrodystrophy, polydactyly, ectodermal dysplasia, and cardiac defects. Although this tetrad constitutes the classical syndrome description, a variable spectrum of clinical manifestations is frequently present. Additional endocardial cushion defects have also been described, including patent ductus arteriosus, ventricular septal defects and ASDs. In one of the patients with EVC syndrome, cardiac defects consisted of CT, ostium-primum, and secundum ASDs. In this patient, the clinical diagnosis of EVC syndrome was made based on the physical and radiographical findings, like polydactyly and cardiac defects.[1] Polydactyly in EVC syndrome is typically seen as post-axial hexadactyly of the hands or in a few cases, the feet. Interestingly, this child also had polydactyly of all the four limbs along with CT and ostium primum septal defect, suggesting the possible diagnosis of a variant of EVC syndrome.

We report this case to document the rare association of CTS, polydactyly and Ostium-primun ASD as an unusual variant of EVC syndrome. We also highlight the common difficulty encountered in the clinical diagnosis of CT in children. Presence of an apical MDM without loud S1 and opening snap, particularly in a child, is a clinical indicator of CT, and echocardiography plays a pivotal role in the diagnosis of this anomaly. Proper corrections, especially in atypical cases, may require accurate determination of intra-atrial anatomy during surgical interventions.

Diagnostic precision in the so-called heritable heart-hand syndromes is essential for the informed clinical evaluation of other family members who may be affected as well. The continuing discovery of new chromosomal abnormalities confirms once again the genetic heterogeneity of the syndrome. Further genetic analyses of these heart-hand syndromes will elucidate the basic mechanisms underlying cardiac morphogenesis and clarify diagnostic and potentially therapeutic approaches to these congenital heart diseases.

REFERENCES

1. Scurlock D, Ostler D, Nguyen A, Wahed A. Ellis-van Creveld syndrome and dyserythropoiesis. Arch Pathol Lab Med 2005; 129:680-2.
2. Antonielli E, Villani M, Pizzuti A, Di Leo M. [Cor triatriatum associated with inter-atrial defect: diagnosis by transesophageal echocardiography]. Minerva Cardioangiol 1993; 41:255-9. Italian.
3. Nagatsu M. [Clinical classification and surgical treatment of cor triatriatum]. Nippon Kyobu Geka Gakkai Zasshi 1992; 40:473-84. Japanese.
4. Goel AK, Saxena A, Kothari SS. Atrioventricular septal defect with cor triatriatum: case report and review of the literature. Pediatr Cardiol 1998; 19:243-5.
5. Varma PK, Warrier G, Ramachandran P, et al. Partial atrioventricular canal defect with cor triatriatum sinister: report of three cases. J Thorac Cardiovasc Surg 2004; 127:572-3.
6. Basson CT, Solomon SD, Weissman B, et al. Genetic heterogeneity of heart-hand syndromes. Circulation 1995; 91:1326-9.
7. Adamopoulos S, Kokkinou S, Parissis JT, Kremastinos DT. New insight into "heart-hand" syndromes: a newly discovered chromosomal abnormality in a family with "heart-hand" syndrome. Int J Cardiol 2004; 97:129-32.