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Case report

Tetralogy of Fallot with Holt-Oram syndrome

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KEYWORDS

Arrhythmias
Atrial septal defect
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ABSTRACT

Holt-Oram syndrome (HOS) is characterised by mild to severe congenital cardiac defects and skeletal abnormalities of the upper limb. This syndrome is also referred to as Hand-Heart syndrome. The most common cardiac disorder is an ostium secundum detected an atrial septal defect (ASD), followed by ventricular septal defect (VSD) and ostium primum ASD. We report a case of HOS with tetralogy of Fallot (TOF). This association is very rare and is hardly reported in the literature.

Introduction

Holt-Oram syndrome (HOS) was first reported in 1960 by Mary Clayton Holt and Samuel Oram, who detected an atrial septal defect (ASD) in members of four generations of a family. Detected an ASD which was associated with a congenital anomaly of the thumbs which lay in the same plane as the fingers.

The most common congenital heart defect is the ostium secundum ASD, seen in 60% of patients with HOS followed by the ventricular septal defect (VSD). The other associated findings include hand malformations and conduction disturbances, originally described by Holt and Oram. The other complex congenital cardiac malformations, like VSD with infundibular pulmonary stenosis, complete atrio-ventricular (AV) canal defect, mitral valve prolapse, hypoplastic left heart syndrome, coarctation of aorta, subaortic stenosis, patent ductus arteriosus, etc. have rarely been reported in HOS.

Recently, we came across a case of hand malformation with cyanotic congenital heart disease, which was later identified as tetralogy of Fallot (TOF). This association is rarely described in patients with HOS.

Case report

A 2-year-old female child presented with fatiguability, bluish discolouration (cyanosis), and growth retardation, since 6 months of age (Figure 1). Patient also had right hand malformation in the form of radial deviation at wrist and cubitus valgus deformity of right upper limb and hypoplastic right

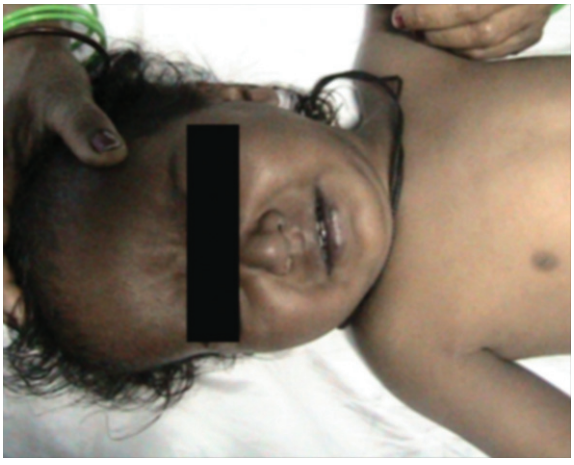

Figure 1 Cyanosis of lips while crying.

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thumb being at the same plane as fingers (distally located than the opposite thumb) (Figure 2). One of her family members had congenital cardiac malformation. Her father's sister had medium size perimembranous VSD, but she had no anomaly related to either of the hands. None of the other family members are affected.

General examination

Patient had cyanosis, which increased on crying and did not improved on 100% oxygen inhalation. Grade II clubbing was also present.

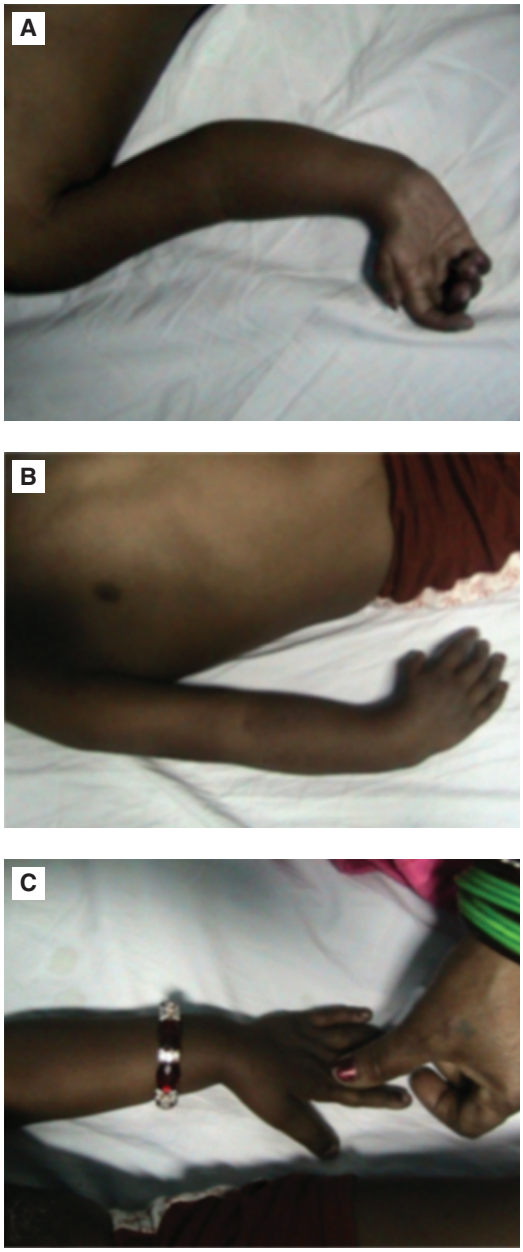

Figure 2 (A, B) Showing right hand deformities (radial deviation of wrist and cubitus valgus deformity of the right elbow). (C) Left hand-deformity.

Cardiovascular system examination

Apex beat was within midclavicular line at the fifth intercostal space. A systolic thrill was present at the upper left intercostal space. Grade I left parasternal heave was present. Ejection systolic murmur was heard at mid and upper left intercostal space. A single second heart sound (S2) was heard.

Musculoskeletal system examination

Right thumb was hypoplastic and located distally in the plane of fingers, in comparison to the left thumb. Right hand was deviated radially at the wrist. Rest of the systemic examination did not reveal any abnormality.

Investigations

Haemoglobin (Hb)—19.8 g/dL, total count—10,100/mm^3, differential count—N40 L52 E3 M5, platelet count—1,14,000/mm^3, haematocrit—60.9%, mean corpuscular volume (MCV)—75.0 fl, red blood cells (RBC) count—$8.13 \times 10^6/mm^3$.

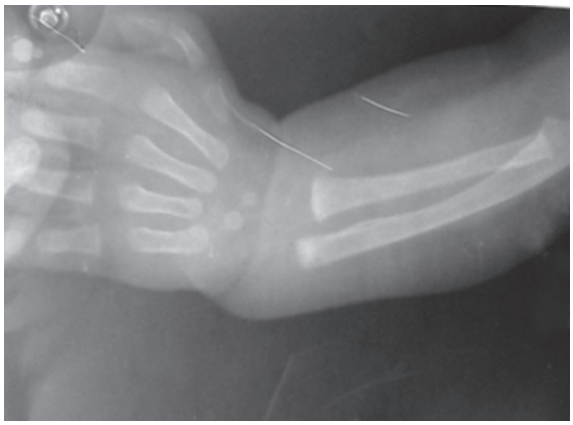

Figure 3 Forearm and hand radiograph showing hypoplastic radius and triphalangeal thumb.

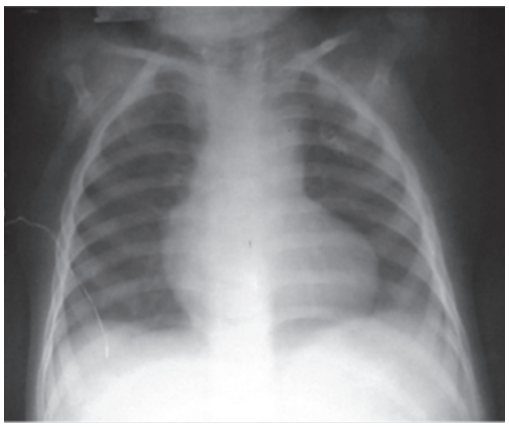

Figure 4 Chest radiograph showing boot-shaped heart and pulmonary oligemia.