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Letter to the Editor

Mirror-image type D interrupted aortic arch: A novel cardiac phenotype providing some perspective in the del22q11.2 syndrome

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Abstract

A 1-day-old baby boy was referred from a local obstetric clinic due to feeble crying, tachypnea, and tachycardia. Two-dimensional echocardiography with Doppler and multidetector computer tomography showed mirror-image type D interrupted aortic arch, conotruncal ventricular septal defect, and patent ductus arteriosus. Emergency cardiac surgery by a biventricular Norwood procedure was performed to relieve his symptom and sign of congestive heart failure successfully. Thymus could not be seen by surgical exploration of the superior mediastinum. Serum C-terminal parathyroid hormone was decreased to less than the normal lower limit. Cytogenetic analysis and fluorescence in situ hybridization study of blood revealed a deletion in chromosome 22q11.2. To the best of our knowledge, mirror-image type D interrupted aortic arch has never been reported in patients with the del22q11.2 syndrome in the English literature. This unusual aortic arch anomaly may provide us a new perspective in the spectrum of cardiovascular malformations in the del22q11.2 syndrome and advocate 22q11.2 deletion as one of the genetic causes of some rare aortic arch anomalies and their correspondent mirror-images.

Keywords: Multidetector computer tomography; Mirror-image; Type D interrupted aortic arch; Conotruncal ventricular septal defect; Biventricular Norwood procedure; Fluorescence in situ hybridization; del22q11.2 syndrome

A 1-day-old baby boy was referred from a local obstetric clinic due to feeble crying, tachypnea, and tachycardia. A single second heart sound was heard over the left upper sternal border. Weak femoral pulse and high-arched palate were noted. Differential cyanosis was noted on the peripheral pulse oximeter, with oxygen saturations of 85% and 65% over the upper and lower extremities, respectively. Plain chest radiograph showed remarkable cardiomegaly. Hypocalcemia was found (total calcium 6.5 mg/dL; ionized calcium 0.91 mmol/L). Two-dimensional echocardiography with Doppler showed ventricular septal defect with conotruncal septal malalignment, right-sided patent ductus arteriosus (PDA), and right aortic arch interruption. Prostaglandin E_1 was administered intravenously at a dosage of 20 ng/kg/min. Multidetector computer tomography (MDCT), which was performed within 24 h of birth, revealed rare aortic arch anomaly, including: 1) interruption of a right aortic arch (denoted by a black star) between the right common carotid artery (RCCA) and the right subclavian artery (RSCA), 2) aberrant left subclavian artery (aLSCA) from the right descending aorta (DAo), and 3) right-sided PDA (denoted by a black asterisk) (Fig. 1). According to the original report of Celoria and Patton, this anomaly is depicted as mirror-image type D interrupted aortic arch (IAA) [1]. Emergency cardiac surgery by means of a biventricular Norwood

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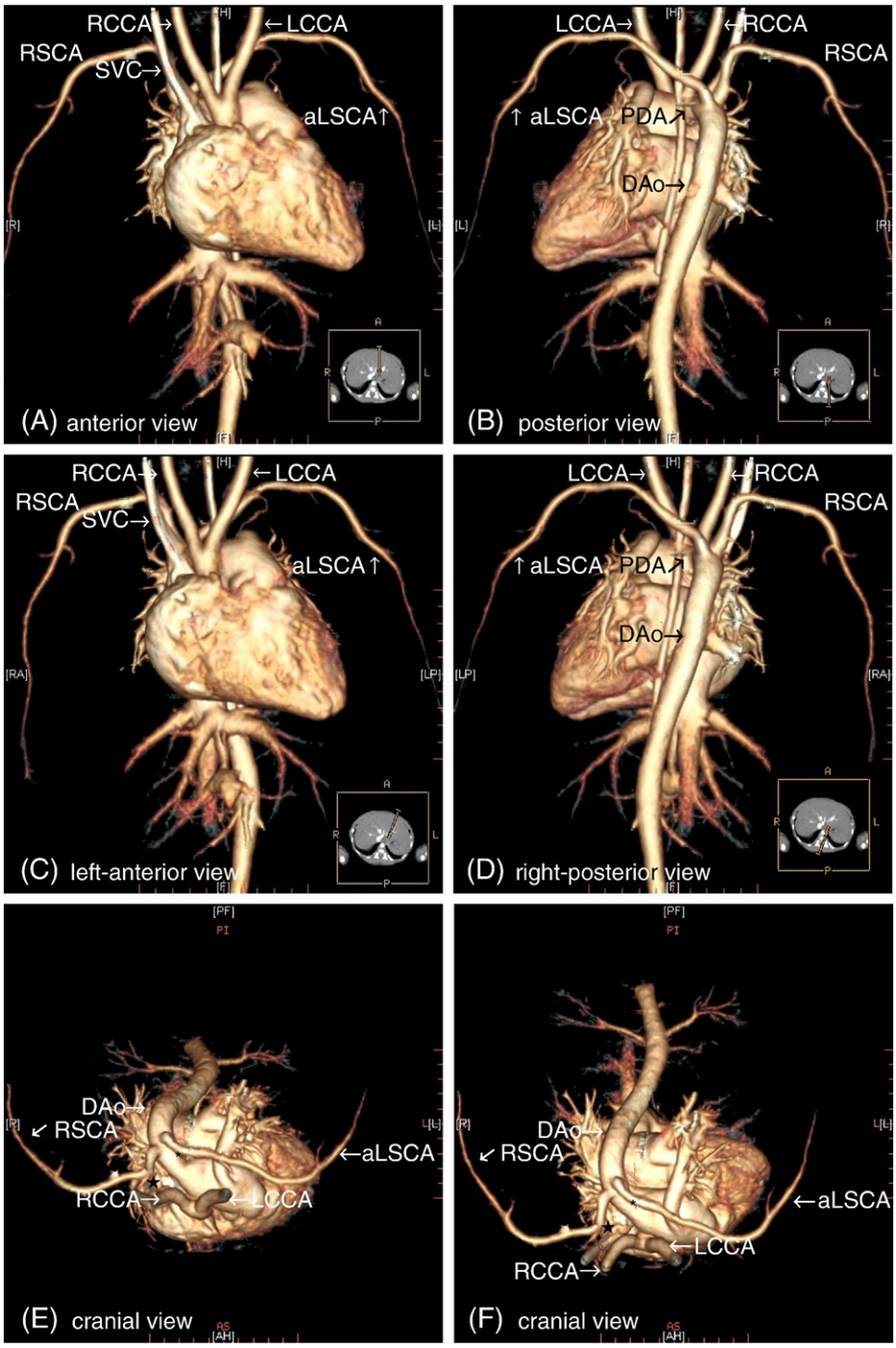

Fig. 1. MDCT showed mirror-image type D interrupted aortic arch.

procedure was intervened to relieve his symptom and sign of congestive heart failure successfully. Meanwhile, thymus could not be found by surgical exploration of the superior mediastinum. Serum level of C-terminal parathyroid hormone was decreased to a level of 0.16 ng/mL (normal range 0.4–1.4 ng/mL). Cytogenetic analysis and fluorescence in situ hybridization study of blood revealed a deletion in chromosome 22q11.2, with a karyotype of 46,XY.ish del(22)(q11.2 q11.2) (TUPLE-1) (Fig. 2).

1. Discussion

The cardiac neural crest cells migrate from the neural fold of the hindbrain to the pharyngeal arches 3, 4, and 6 to play their important roles in the formation of the pharyngeal apparatus and related structures and the outflow tract [2]. Early changes of cardiovascular development can be induced by neural crest ablation in the chick embryo to cause a diversity of cardiovascular anomalies, including cardiac inflow anomalies,