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Brief Report

Atrioventricular septal defect with common atrioventricular junction, common arterial trunk, and severe coarctation of the aorta in a patient with Down's syndrome

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Abstract Atrioventricular septal defect with common atrioventricular junction is rarely associated with either a common arterial trunk or coarctation of the aorta. We report, as far as we are aware, the first case of Down's syndrome with atrioventricular septal defect with common atrioventricular junction, common arterial trunk, and severe coarctation of the aorta.

Keywords: Congenital heart disease; genetic syndrome; Trisomy 21

Atrioventricular septal defect with common atrioventricular junction is common in patients with Down's syndrome, but is rarely associated with either common arterial trunk or coarctation of the aorta. We report a patient with all these lesions, an unusual combination of defects which, as far as we are aware, has not previously been reported.

Case report

A full-term male neonate was born to a 32-year-old primigravida by spontaneous vaginal delivery. The antenatal history was significant for the diagnosis of Down's syndrome. Fetal echocardiography had revealed an atrioventricular septal defect with common atrioventricular junction together with a common arterial trunk. There was no history of exposure to radiation, drugs or alcohol during the pregnancy. Neither was there any significant family history for cardiac defects. The baby was delivered in a tertiary hospital weighing 2580 grams and required no resuscitation.

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On physical examination, the baby appeared dusky but was haemodynamically stable, with pulse oximetry of 87% in room air. The baby had features consistent with Down's syndrome. On cardiovascular examination, the first heart sound was normally heard. The second heart sound was single. There was evidence of a midsystolic ejection click. No cardiac murmurs were appreciated at the time of the initial examination. All peripheral pulses were well felt. The baby was breathing comfortably and the respiratory examination was unremarkable. The remainder of the examination was unremarkable.

Chest radiograph revealed mild cardiomegaly with normal pulmonary vascularity. Capillary blood gas was within normal limits with no evidence of acidosis. A cross-sectional echocardiogram was performed, revealing a common atrioventricular valve of Rastelli type. A pattern, a left dominant atrioventricular septal defect with common atrioventricular junction, and with mild atrioventricular valvar insufficiency. There was also evidence of a common arterial trunk, with mild to moderate truncal valvar insufficiency. The aortic arch was left-sided, with hypoplasia of the transverse arch and severe coarctation of the aorta. A large patent arterial duct was present, with bidirectional shunting. The ventricular function appeared normal (Figs 1, 2a and 2b). The baby was started on prostaglandins. Karyotypic examination revealed trisomy 21, and fluorescent in-situ hybridization for 22q11 deletion was negative.

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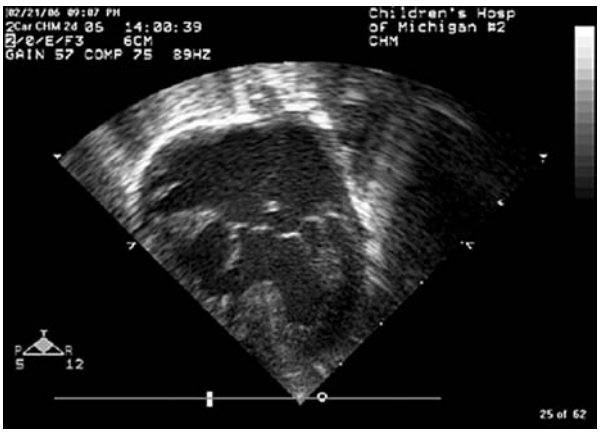

Figure 1.
Atrioventricular septal defect with common atrioventricular junction with hypoplasia of the right ventricle.

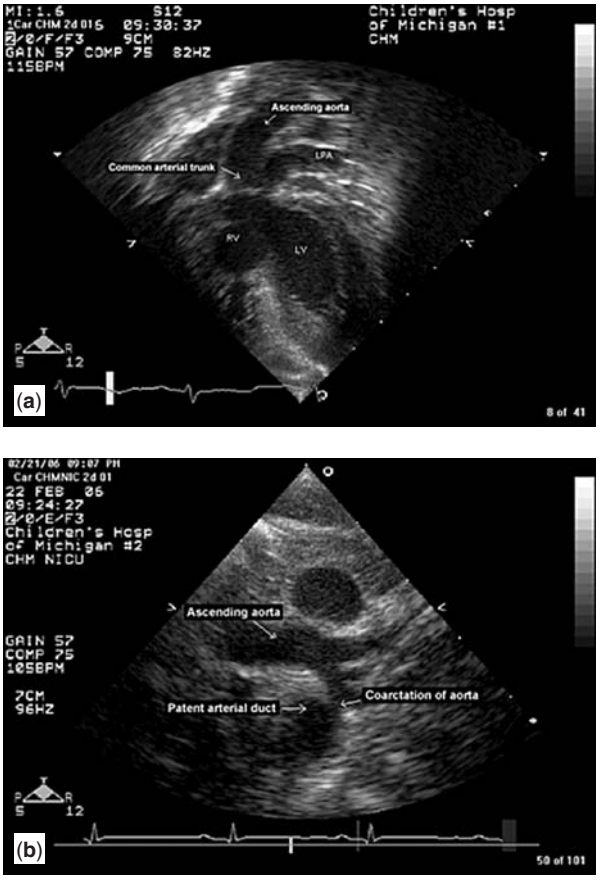

Figure 2.
*Evidence (**a**) of common arterial trunk with the pulmonary arteries arising from the common trunk. There is also evidence (**b**) of hypoplastic transverse aortic arch and severe coarctation of the aorta.*

In view of the anatomy in presence of Down's syndrome, the parents were given options ranging from no intervention to surgical palliation and cardiac transplantation. The parents opted for no intervention. The prostaglandin infusion was stopped, and the baby was discharged home under hospice care.

Discussion

Atrioventricular septal defect with common atrioventricular junction with common arterial trunk is a rare combination of defects. Sousa-Uva et al.,[1] in their case report of a successful repair of a patient with this combination, noted that there had been only 11 previously reported cases of similar anatomy. Arai et al.[2] subsequently have reported one more case. Collett and Edwards[3] reported 19 specimens of common arterial trunk with atrioventricular septal defect with common atrioventricular junction. Van Praagh,[4] however, denied this association in a later review of the same specimens. None of the reported cases were associated with arch anomalies.

Atrioventricular septal defect with common atrioventricular junction is rarely associated with coarctation of the aorta. In an autopsy series of 38 such patients, Campbell et al.[5] reported 3 out of 38 to have coarctation of the aorta. Redmond et al.[6] reported 100 such patients undergoing cardiac surgery. They found only 1 patient to have coarctation of the aorta, and 1 patient was reported to have hypoplasia of the aortic arch.

There have been occasional case reports of coarctation of aorta in patients with Down's syndrome. Detailed search revealed a total of 12 patients of coarctation of the aorta out of a total of 950 patients with Down's syndrome in different case series, with an estimated prevalence of 1.26%. In all the patients from the series of patients with Down's syndrome and coarctation of the aorta, there was no reported patient with common arterial trunk. Our patient, therefore, is the first to be reported with Down's syndrome associated with this unusual combination of lesions.

Sousa-Uva et al.[1] reported a successful repair of a patient with balanced atrioventricular septal defect with common atrioventricular junction with common arterial trunk. Trowitzsch et al.[7] reported an unsuccessful attempt at surgical correction of a similar lesion. Formigari et al.[8] reported favorable outcomes for patients with Down's syndrome with atrioventricular septal defect with common atrioventricular junction amenable to biventricular repair but only one of six patients undergoing single ventricle repair survived to Fontan palliation. In our case, the parents were given the options of surgical palliation with functionally univentricular physiology, heart transplantation, or no intervention. They opted for the latter alternative, and therefore no surgical procedure was performed.

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