

Letter to the Editor

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

Sir,

We read the interesting paper by Singh and Pettersen¹ reporting on a patient with Down's syndrome with atrioventricular septal defect and common atrioventricular junction, common arterial trunk and aortic coarctation. The association between atrioventricular septal defect with common arterial trunk and usual atrial arrangement is very rare.² Because of this, therefore, we report here on two interesting cases recently observed by our own group.

The first patient was a nonsyndromic neonate with atrioventricular septal defect with common atrioventricular valve and common arterial trunk with a confluent pulmonary arterial segment, the so-called "type 1". Interestingly, this patient was the first cousin of a nonsyndromic child with interruption of the aortic arch between the left common carotid and subclavian arteries, the "type B" variant.³ The second patient had Down's syndrome with common arterial trunk and interrupted aortic arch, who presented also a "cleft" of the otherwise normal mitral valve.⁴

A common arterial trunk is extremely rare in patients with Down's syndrome, but in both our second case³ and in the case reported by Singh and Pettersen,¹ this malformation was associated with obstruction in the aortic arch, in spite of

the fact that isolated aortic coarctation or interruption is very rare in the setting of trisomy 21. All these observations confirm the mysterious interrelations that still exist between genetics, familial recurrence and complex congenital cardiac defects.

*Claudia Saffirio, Anna Chiara Vittucci
Bruno Marino
Pediatric Cardiology
Department of Pediatrics
University of Rome
Italy*

References

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The letter was shown to Drs Singh and Pettersen, who commented that they had nothing further to add.

Correspondence to: Prof. Bruno Marino, MD, Pediatric Cardiology, University of Rome "La Sapienza", Viale Regina Elena 324, 00161 Rome, Italy. Tel: +39 06 49979210; Fax: +39 06 49970356; E-mail: bruno.marino@uniroma1.it