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Prevalence of congenital heart defects in patients with Down's syndrome

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Down's syndrome (DS) is the most frequent chromosomal aberration among newborns, with an incidence of 1/660 live births; trisomy 21 occurs in 95% of cases due to maternal meiosis I non-disjunction, resulting in three full copies of chromosome 21 in each cell. Of these cases, 4% are due to parental/de novo translocation and 1% is due to mosaicism. Frequency of congenital heart defects (CHDs) in children with DS varies greatly in the literature, from 20 to over 60%.^{1,2}

In this issue of *Jornal de Pediatria*, Vilas Boas et al.³ publish a study aimed at determining the prevalence of CHDs in patients with DS in the municipality of Pelotas, Brazil, describing the most frequent types and assessing associated factors. The authors' cross-sectional study included children with DS who were born and lived in Pelotas from January 2000 to December 2005. Data were collected by means of home interviews with mothers or guardians. There is no surprise in their findings with regard to the prevalence of DS and CHD in DS.

Bivariate analysis between the outcome CHD and the predicting factors maternal age, paternal age, parents' and child's skin color, presence of other malformations,

and child's sex showed that the associations were not statistically significant. These findings correlated with 532 affected children found in the Atlanta project.⁴

The most frequent heart defect in the study by Vilas Boas et al.³ was interatrial communication (17%); atrioventricular septal defect (AVSD) affected five patients. Complete AVSDs

are one of the most common cardiac defects in DS, but the distribution of CHDs in children with DS may vary according to geographical location.^{5,6} In epidemiological studies carried out in the United States and Europe, a complete form of AVSD reached the highest rate, affecting up to 60% of patients.^{7,8} Alternately, in Asia, isolated

ventricular septal defects have been reported to be the most common defects, observed in about 40% of patients.⁹ In Latin America, a secundum type of atrial septal defect (ASD) was reported to be the most common lesion (40%).⁵ This data correlated with Latin American studies.

Age at evaluation of CHD was low in the study by Vilas Boas et al.³: 63.8% of the patients were evaluated during the first 6 months of age, and most of them had echocardiograms (93.6%). This striking finding is the

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No conflicts of interest declared concerning the publication of this editorial.

Suggested citation: Nisli K. Prevalence of congenital heart defects in patients with Down's syndrome. *J Pediatr (Rio J)*. 2009;85(5):377-378.

doi:10.2223/JPED.1940

most important factor for the reduction of mortality and morbidity, because irreversible pulmonary vascular disease developing early in DS with left-to-right shunt lesions are related exactly to AVSDs. Morris et al.¹⁰ reported that the operative outcomes of children with DS and complete AVSDs are highly correlated, with early progression to obstructive pulmonary vascular disease in these patients being one of the most important factors because of its high perioperative mortality. There are also arguments about the impact of the association of DS on the clinical outcomes of surgical repair of complete AVSDs, including late mortality and morbidity.¹¹ However, Rizzoli et al.¹² reported that DS was not an independent risk factor for operative mortality and that patients with DS underwent fewer reoperations. In spite of some conflictive points between the two groups, we do suggest that in patients with DS and AVSDs, surgery should be performed before 12 months of age, preferably at 3 to 6 months of life and before the onset of pulmonary hypertension.

Vilas Boas et al.³ also report a very low percentage of patients submitted to karyotype testing (49%) and an even lower percentage of prenatal diagnosis (2.1%). Antenatal genetic counseling in cases with a fetal echocardiographic diagnosis of complete AVSDs is mandatory due to its strict association with DS and other chromosomal anomalies. At present, prenatal diagnosis of complete AVSDs has been associated with a 58% risk of aneuploidy, mainly DS.¹³

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