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Letters to the Editor

Types of cardiac defects in children with Down's syndrome

Sir,

We thank Placidi, Digilio, and Marino for their interest in our work. Our findings concerning the principal association between patency of the arterial duct and Down's syndrome in Guatemala are clearly in contrast with the classic epidemiologic reports, where atrioventricular septal defects with common atrioventricular junction, and ventricular septal defects, are the lesions most frequently observed in Caucasian[1,2] and Asian[3,4] populations with Down's syndrome. Our data,[5] nevertheless, is similar to that reported from other Latin-American countries. In Mexico,[6] for example, patency of the arterial duct was also found to be the most frequently associated lesion in patients with Down's syndrome. Our data is also in keeping with that from Mexico[6] with regard to the frequency of ventricular and atrioventricular septal defects, which proved to be the second and the fourth most frequent isolated cardiac malformations in such patients.

The cardiac phenotype is, indeed, the result of many genetic interactions and specific embryological mechanisms, especially in children with major genetic defects.[7] Ethnic and geographic factors, nonetheless, may also influence the formation of these anomalies. Guatemala has close to 13 million inhabitants, of whom approximately nine-tenths are of Mayan descent and Ladinos, while the remaining one-tenth of the population is made up of minority racial groups, such as Whites, Garifunas, and Xincas. About two-fifths of the population is extremely poor, earning less than 2 United States dollars each day, and have very limited access to medical care. In our paediatric cardiac surgical unit, two-thirds of children with congenital cardiac malformations, including those with Down's syndrome, also suffered from chronic malnutrition. Furthermore, two-thirds of the patients also live at least 1500 metres above sea level.[5] Conceivably, all these ethnic-geographic factors may also contribute to explain the different frequency of associated cardiac malformations seen in our population with Down's syndrome. Further genetic studies are clearly essential to explain the encountered differences in cardiac phenotype in the various geographic populations with Down's syndrome.

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The Letter was also shown to Dr Samson and his colleagues, who thank the group from Bambino Gesù Hospital for their comments, with which they agree.

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Types of cardiac defects in children with Down's syndrome

Sir,

In recent issues of your journal, we read the interesting papers by Samson and Kumar[1], and by Vida et al.[2] These studies, although concerning different topics, provide interesting data regarding the prevalence of congenital cardiac malformations in patients with Down's syndrome. Samson and Kumar[1] reported that, in their neonatal population with Down's syndrome from Arab Emirates, "ventricular septal defects and atrial septal defects in the oval fossa are the most common malformations". In contrast, Vida et al.[2] reported that the most frequent malformation in patients with Down's syndrome in Guatemala is patency of the arterial duct, albeit followed by ventricular septal defect.

These findings are in contrast with the classic epidemiological papers on cardiac disease in patients with Down's syndrome, which show atrioventricular septal defect with common atrioventricular junctions to be the most common congenital cardiac malformation in Caucasian populations.[3]

It is of interest to note, therefore, that also in Chinese[4] and Japanese[5,6] populations, the most frequent cardiac malformation in patients with Down's syndrome is not an atrioventricular septal defect with common junction, but a ventricular septal defect. Mexican data[7] is similar to the oriental findings, because the most common cardiac defect is again ventricular septal defect, followed by atrioventricular septal defect. It would be of interest, of course, to know whether any of those reported with ventricular septal defect also had common atrioventricular junctions.

A definitive explanation for this different prevalence of congenital cardiac malformations in children with Down's syndrome, however, remains unknown.

There are several hypotheses. The authors from Arabian countries suggest the possible role of the time of the screening. The high number of ventricular septal defects found in their study could be due to the neonatal age of their patients, since some ventricular and atrial communications could close spontaneously with time. Another explanation for the prevalence of patency of the arterial duct in patients with Down's syndrome from Mexico, Perù and Colombia, as proposed by Vida et al.,[2] is the lower partial pressure of oxygen at high altitude. Other reports[7,8] have shown this to be a determinant of patency of the duct.

These interesting hypotheses, however, cannot explain the data from oriental and Mexican populations[4–7] and their difference from the Caucasian series.[3] Different prevalence of specific cardiac malformations amongst ethnic groups was previously reported and explained on the basis of genetic heterogeneity. For example, the subarterial ventricular septal defect is more frequent in the setting of tetralogy of Fallot in the Japanese population[9] than in Caucasian populations.[10]

The cardiac phenotype is the result of many genetic and environmental interactions. In children with Down's syndrome, furthermore, additional genetic[11,12] and/or environmental[13] variants may contribute to cardiac morphogenesis. We might anticipate, therefore, that ethnic genetic factors could also change the prevalence of types of cardiac defects in children with major genetic defects.[14] If this were the case, it could help to explain the incomplete correlations between genotype and phenotype, and the significant phenotypic variability observed in patients with genetic syndromes. It could also prompt a search for additional modifying genetic or environmental factors involved in cardiac morphogenesis.

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Sir,

Re: Intervention for overweight and obese school children

The intention of Graf and her colleagues[1] was to prove the value of early preventive measures in obese children. Values for systolic blood pressure were higher in obese children compared to those of normal body weight. Intervention was shown to have a significant effect on systolic blood pressure. The study, however, fails to delineate specific effects in the subgroup of obese children with hypertension. It is not clear, indeed, whether the intervention had a positive effect.

The majority of obese children was normotensive, and the statistical mean of systolic blood pressure was normal for the whole group. Since obese children tend to be taller, it is no wonder that their blood pressure was higher. We presume that a maximum of 4 children amongst the 40 undergoing intervention were hypertensive. What effect did the intervention have on these 4? Most probably this particular group was too small for statistical analysis. The whole group showed a regression to the overall mean within the cohort of normotensive values, which is a well known statistical phenomenon.

Since the study outlines expensive measures for health care, it should be continued, hopefully to clarify whether hypertension in obese children can indeed be treated successfully by the proposed intervention.

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Sir,

On behalf of my colleagues, I thank Dr Lehn for his letter. We are delighted with the interest he has shown in our work. Because of the importance of his question, we have calculated the effect of intervention in the group of obese and overweight children. Of 35 children undergoing intervention, 5 were classified as hypertensive, accepting the limitation that their blood pressure was measured only once. After intervention, only 1 child remained hypertensive. No significant differences were found compared with the control group.

The correlation in the group of children undergoing intervention and their controls showed a weak significant correlation between reduction of the body mass index and reduction of diastolic blood pressure ($r = 0.214$; $p = 0.005$), but not the systolic blood pressure. After separating the two groups, the positive correlation was stronger, and remained only found in the group undergoing intervention ($r = 0.518$; $p = 0.001$ versus $r = 0.105$; $p = 0.232$ in the controls).

Our purpose in the study reported was to demonstrate the effect of the StEP Two programme on blood pressure in general. It is inappropriate to draw further conclusions at this stage. Serial and systematic follow-up data is warranted to define the effect on obese and hypertensive children.

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