Cardiac Defects in Mexican Children With Down Syndrome

To the Editor,

It was with great interest that we read the recently published artic1e by de Rubens Figueroa et al[1] in REVISTA ESPAÑOLA DE CARDIOLOGÍA. The article contained a report that the most frequently observed isolated cardiac defects in Mexican children with Down syndrome are atrial septal defects and ventricular septal defects. This observation, which has been described previously,[2] contrasts with reports of the types of cardiac defect found to be prevalent in Caucasian children with Down syndrome. In these latter children, persistence of the atrioventricular canal is the most common heart malformation.[3-5]

It is interesting to note that the most frequently occurring cardiac defect in Chinese[6] and Japanese[7,8] patients with Down syndrome is not persistence of the atrioventricular canal but ventricular septal defect. Perhaps the genetic similarities between oriental and native American populations that have been reported[9] could cause the similar prevalence of these different types of cardiac defect.

However, variations in the prevalence of specific cardiac malformations in children with Down syndrome from different ethnic backgrounds have been noted before[10,11] and may be explained by heterotrisomy of chromosome 21,[12] by genetic variations in some chromosome other than chromosome 21, or by environmental factors.

Nevertheless, de Rubens Figueroa et al's paper clearly confirms that, even in the presence of a major genetic anomaly, the phenotype can be influenced by numerous genetic and environmental factors.

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Response

To the Editor,

We are grateful to Dr Marino and colleagues for their comments on our recent publication in REVISTA ESPAÑOLA DE CARDIOLOGÍA. It is certainly interesting to draw attention to the fact that the same well-studied chromosomal alteration, such as the one responsible for Down syndrome (which is also the commonest), can, in various parts of the world, result in a range of different heart conditions, which may even stem from distinct embryological defects. These defects can be in interatrial or interventricular septum development (which can result in interatrial or interventricular communications) or in atrioventricular septum formation (which can result in a persistent atrioventricular canal, thereby possibly disturbing tricuspid or mitral valve development).

In addition, Dr Marino and colleagues also point out the curious similarities between the heart conditions observed in Down syndrome children in Asia and in Latin American, including Mexico. It is interesting to note that studies of genetic markers in different races show that Mexicans are very similar to Asians. Perhaps this is due to the fact that our ancestors reached the American continent by crossing the Bering Strait from Asia, after which they became established here. The same process did not occur in the United States or Canada, which were populated many years later by Anglo-Saxons and whose populations therefore derived a genetic similarity with Europeans.

It is highly significant that environmental factors present in different geographical regions can also influence the way in which genetic diseases such as heterotrisomy manifest themselves. This phenomenon supports the view that many illnesses develop because of the combination of genetic factors with environmental and behavioral triggers, as was suggested some time ago. Nevertheless, the fact that the specific way in which common chromosomal illnesses manifest themselves also comes under external influence means that a range of causal factors are involved and that, consequently, there may be a range of novel approaches to therapy.

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