Heterotaxia syndromes and 22q11 deletion

In a recent issue of your journal we read with interest the very accurate review by Penman Split et al. on defects of left-right asymmetry. The authors correctly reported that in patients with heterotaxia (asplenia and polysplenia syndromes), conotruncal defects are one of the more frequent heart malformations. It is well known that 22q11 deletion has been described in a subgroup of patients with conotruncal anomalies in the setting of Di-George\(^4\) and velocardiofacial syndromes. In the paper of Penman Split et al.\(^3\) was reported (personal communication to the authors) that the same microdeletion has been found in two patients, one with dextrocardia and one with left isomerism (polysplenia syndrome).

Since 1993 we have performed clinical and molecular evaluation of all patients with conotruncal anomalies observed at our hospital,\(^1\)\(^2\) including 20 cases with heterotaxia. Fifteen had asplenia syndrome and five polysplenia. All patients underwent phonotypic and cardiac examinations. Fluorescent in situ hybridisation was used for detecting 22q11 deletion.

No patients had the phenotypic features of Di-George\(^4\) or velocardiofacial syndromes, and the genetic study did not show 22q11 deletion in any case. Our experience suggests that the conotruncal anomalies in the setting of heterotaxy syndromes are not related to 22q11 deletion, and are probably secondary to distortion of cardiac looping or to the anomaly of the situs itself. Different gene(s) and different developmental mechanisms may be involved in the pathogenesis of conotruncal anomalies in patients with situs solitus and in those with laterality defects.

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