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*Letter to the Editor*

# Deficiency of the infundibular septum in patients with Interrupted aortic arch and del 22q11

WE READ WITH INTEREST THE PAPER OF Momma et al. concerning interruption of the aortic arch in association with deletion of chromosome 22q11.[1]

In 6 of the 7 patients with this association the authors reported 'a complete deficiency of the muscular outlet septum with the defect extending to the perimembranous area'. Since the subarterial and doubly committed ventricular septal defect is prevalent in the oriental populations,[2] Momma et al. suggest that 'It remains to be confirmed if the association of the specific type of defect seen with interrupted aortic arch between the left common carotid and the left subclavian arteries and deletion of chromosome 22q11 is present in Caucasians'.

Contemporany to the paper of Momma et al.,[1] we published a study on the same topic involving analysis of Italian patients.[3] Among 13 children with interruption of the aortic arch between the common carotid and subclavian arteries, and with deletion of chromosome 22q11, we detected in 9 patients the same pattern of ventricular septal defect, in which there was complete absence of the muscular outlet septum. We can conclude that this type of ventricular septal defect, previously reported in patients with interrupted aortic arch,[4,5] is characteristic of the specific combination of the site of interruption and deletion of chromosome 22q11 in spite of the racial differences.

Hypoplasia or absence of the muscular outlet septum has also been reported to be prevalent in patients with tetralogy of Fallot and deletion of chromosome 22q11[6,7] suggesting that a specific disturbance of the infundibular musculature is very frequent in this genetic syndrome.[8] In fact, the muscular outlet septum is hypoplastic or absent in other malformations of the ventricular outflow tracts associated with deletion of chromosome 22q11, including common arterial trunk,[9] tetralogy with pulmonary atresia,[10] and in some patients with isolated ventricular septal defects (unpublished observation).

Recently, Yamagishi et al.,[11] studying the expression of a new gene that our group previously identified,[12] reported that 'UFD1L expression was most evident in the fourth aortic arch artery which is responsible for formation of the segment of the aortic arch that lies between the left carotid and subclavian arteries'.

As pointed out in the editorial comment of McElhinney and Anderson,[13] and in agreement with these new anatomic and genetic observations, we suggest that in cases where the aortic arch is interrupted between the left carotid and subclavian arteries the hypothesis of a pathogenetic role of subaortic obstruction[5–14] has lost value.

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