

Letter to the Editor

Right aortic arch with coarctation proximal to the right subclavian artery and Kommerell's diverticulum

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TSUBATA ET AL.¹ RECENTLY DESCRIBED AN interesting case with right aortic arch and coarctation proximal to the right subclavian artery. Right aortic arch is rare in patients with aortic coarctation, and was not present in the 154 patients in our earlier study.² Recently, however, we have reported the congenital malformations observed in association with chromosome 22q11 deletion.³ One of the 100 patients studied had almost exactly the same complex as reported by Tsubata et al.¹ Our case had normal intracardiac anatomy, right aortic arch, coarctation proximal to the right subclavian artery producing a pressure gradient of 50 mmHg, aneurysmal dilation of the proximal part of the right subclavian artery, a retroesophageal arch providing aberrant origin of the left subclavian artery, and Kommerell's diverticulum. Chromosome 22q11 deletion is characteristically associated with anomalies of the aortic arch, subclavian artery, arterial duct and pulmonary arteries, either associated with tetralogy of Fallot,^{4,5} common arterial trunk⁶ or a normal heart.³ The patient described by Tsubata et al.¹ should be checked for chromosome 22q11 deletion. DiGeorge syndrome and chromosome 22q11 deletion are frequently associated with interruption of the aortic arch between the common carotid and left subclavian arteries, and almost never with interruption at the isthmus.⁷ This suggests that the aortic arch proximal to the left subclavian artery is a specific developmental weak point, and tends to be stenotic or interrupted in some patients with chromosome 22q11 deletion.

References

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The letter of Dr Momma was shown to Dr Tsubata and colleagues, who responded as follows:

We thank Professor Kazuo Momma for his comments concerning our description of 'Balloon angioplasty for isolated coarctation of the aorta with mirror image right aortic arch in an infant'.¹ According to his suggestion, we performed the fluorescence in situ hybridization analysis in our patient with right aortic arch and coarctation. We could detect no chromosome 22q11 deletion. We also investigated the chromosomes for 22q11 deletion simultaneously in a recent case with tetralogy of Fallot, pulmonary atresia, and conotruncal anomaly face as a control. Chromosome 22q11 deletion proved to be present only in this control. Our case had right aortic arch and coarctation proximal to the right subclavian artery, almost exactly as reported by Momma et al.² In our case, however, we did not identify any facial anomalies.

In the elegant study of Momma et al,² in contrast, all patients with the deletion showed the so-called 'conotruncal anomaly face'. In contrast, none of their 117 patients with heart disease, but without 'conotruncal anomaly face', had the deletion. Their fascinating findings indicate that the combination of cardiac and facial anomalies is essential in the clinical recognition of the 22q11 deletion. Our patient has shown satisfactory growth and development without any symptomatology referable to DiGeorge syndrome or CATCH 22 syndrome. Thus, our patient may be a rare example of the combination of right aortic arch and coarctation not associated with chromosome 22q11 deletion.

References

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