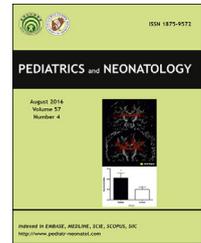


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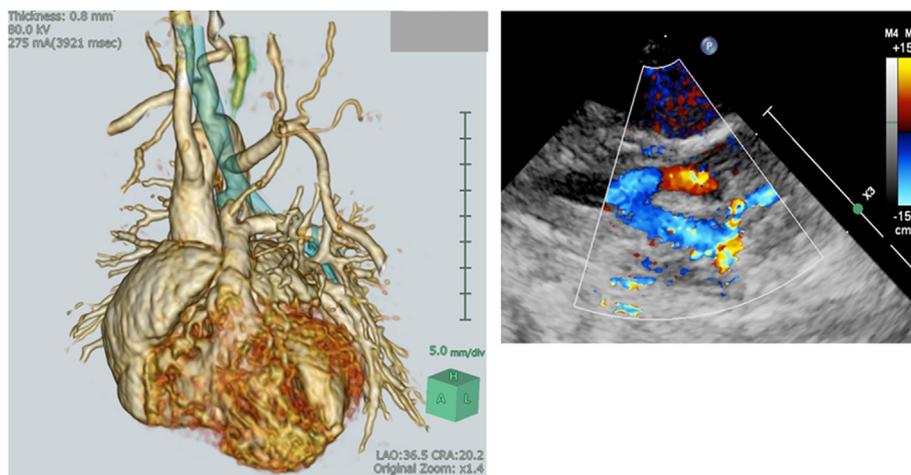
Letter to the Editor

## 22q11.2 deletion syndrome comorbid with tetralogy of fallot and left common carotid artery congenital aplasia: A case report

The 22q11.2 deletion syndrome concomitantly occurs with cardiovascular abnormalities in 80% of all cases, for which three genes on chromosome 22q11.2 (*TBX1*, *CRKL*, and *ERK2*) were responsible. Common carotid artery (CCA) congenital aplasia is a rare vascular malformation. Herein, we report for the first time a case of a male infant with 22q11.2 deletion syndrome comorbid with tetralogy of Fallot (TOF) and left CCA congenital aplasia.

A male infant was vaginally delivered at 40 weeks and 1 day of gestation with a birth weight of 2952 g and a length of 49.7 cm. He was the first-born child with unremarkable family history. His heart rate was 120 beats per minute, blood pressure was 64/35 mmHg, respiratory rate was 40 breaths per minute, and oxygen saturation was 92%. No

external deformities were noted. Chest X-ray revealed a cardiothoracic ratio of 62.5% and a heart shadow that resembled a wooden shoe. Laboratory findings revealed no electrolyte abnormalities. Blood gas analysis showed no hypoglycemia or acidosis. Postnatal echocardiography revealed TOF with the right aortic arch (RAA). Contrast-enhanced computed tomography showed that the right CCA, right subclavian artery, and aberrant left subclavian artery branched off from his RAA, in order, without left CCA branching. The right and left vertebral arteries branched from each subclavian artery, respectively. Additionally, Kommerell's diverticulum was not observed. The blood flow direction of the left internal and external carotid arteries was opposite on echocardiography (Fig. 1). Magnetic



**Figure 1** Enhanced computed tomography and color Doppler imaging of echocardiography. Enhanced computed tomography shows congenital aplasia of the left common carotid. Echocardiography shows retrograde blood flow from the left internal carotid artery to the left external carotid artery.

<https://doi.org/10.1016/j.pedneo.2022.05.017>

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Please cite this article as: K. Tao, J. Hashimoto, Y. Ishikawa et al., 22q11.2 deletion syndrome comorbid with tetralogy of fallot and left common carotid artery congenital aplasia: A case report, *Pediatrics and Neonatology*, <https://doi.org/10.1016/j.pedneo.2022.05.017>

resonance angiography revealed no right posterior communicating artery in the circle of Willis, as well as atrophy and ischemic findings in the left cerebral hemisphere.

Completely absent ipsilateral CCA and vertebral artery with contralateral internal carotid artery aneurysms were reported in some adult cases, but the branching morphology of RAA is different from our patient.<sup>1</sup> Additionally, our patient did not have cerebral aneurysms, which were reported in 13.7% of cases with aplasia of the CCA.<sup>2</sup> One report of an infant case, involving the RAA in 22q11.2 deletion syndrome comorbid with a ventricular septal defect, has been published.<sup>3</sup> In that case, the left CCA branched from the left pulmonary artery and showed retrograde blood flow via the circle of Willis, but the ductus arteriosus with RAA generally originates from the left subclavian artery, as in our patient. After closing the ductus arteriosus in our patient at 3 days, his oxygen saturation was maintained at 94%–97%, without changes in other vital signs. The diagnosis of the 22q11.2 deletion syndrome was genetically confirmed with parental consent. RAA was defective in the left third pharyngeal arch in the embryo, suggesting that blood flow to the left cerebral hemisphere was maintained through the circle of Willis since fetal development.

### Declaration of competing interest

The authors have no conflict of interest to declare.

### Acknowledgements

This research was not supported by any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

### Appendix A. Supplementary data

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.pedneo.2022.05.017>.

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Katsuo Tao\*

Department of Cardiology, Japan

Joji Hashimoto

Department of Radiology, Fukuoka Children's Hospital,  
Japan

Yuichi Ishikawa

Kouichi Sagawa

Department of Cardiology, Japan

\*Corresponding author. 5-1-1, Kashiiteriha, Higasi-ku,  
Fukuoka city, Fukuoka, 813-0017, Japan.  
E-mail address: [katsuo1203@yahoo.co.jp](mailto:katsuo1203@yahoo.co.jp) (K. Tao)