A rare cause of neck swelling in a child

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DESCRIPTION

A 6-year-old boy who presented with sore throat had bilateral swellings in the submandibular regions (figure 1A). Surprisingly the swellings were pulsatile. Detailed history revealed generalised hypotonia during infancy with motor developmental delay. He had undergone sternotomy for closure of a large atrial septal defect at the age of 4 years. On examination he had lean build with long face, high arched palate, large ears, sternotomy scar on the chest (figure 1B) and bilateral inguinal herniae. There was mild increase in skin elasticity but no joint hyperextensibility. There was no cleft palate or bifid uvula. Family history revealed exactly similar features in the younger brother. MR angiogram of neck showed multiple tortuosities of bilateral common carotid and bilateral vertebral arteries (figure 1C).

Arterial tortuosity syndrome and Loyes-Dietz syndrome are the closest differential diagnoses for this child. Arterial tortuosity syndrome occurs due to mutation in the gene (SLC2A10) encoding for glucose transporter GLUT10 whereas Loyes-Dietz syndrome occurs due to mutation in transforming growth factor β-receptor (TGFBR) 1 or 2. Differentiation of these two conditions only on the basis of clinical grounds is not possible and requires genetic analysis for mutations in SLC2A10, TGFBR1 and TGFBR2. However, we could not obtain genetic analysis of our patient.

The most common vascular swelling in the neck of children is an aneurysm due to congenital, mycotic or post-traumatic reasons. As in our case, arterial tortuosity could be rare causes of vascular swellings in neck of children.

Learning points

▸ Arterial tortuosity could be a rare cause of vascular swelling in the neck of children.
▸ It is very essential to identify the vascular swellings in children to avoid disastrous attempts of aspiration or biopsy.

Competing interests None.

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REFERENCES