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 built 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 Loeys-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 Loeys-Dietz syndrome occurs due to mutation in transforming growth factor β -receptor (TGFB1) 1 or 2.^{1 2} Differentiation of these two conditions only on the basis of clinical grounds is not possible and requires genetic analysis for mutations in SLC2A10, TGFB1 and TGFB2. 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.³ As in our case, arterial tortuosities could be rare causes of vascular swellings in neck of children.

Figure 1 Clinical photograph of the patient showing right-sided neck swelling (A) with long slender face and sternotomy scar (B) and MR angiogram of major neck vessels showing marked tortuosities of bilateral carotid and vertebral arteries (C).

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.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Ritelli M, Drera B, Vicchio M, *et al*. Arterial tortuosity syndrome in two Italian paediatric patients. *Orphanet J Rare Dis* 2009;4:20.
- 2 Drera B, Ritelli M, Zoppi N, *et al*. Loeys-Dietz syndrome type I and type II: clinical findings and novel mutations in two Italian patients. *Orphanet J Rare Dis* 2009;4:24.
- 3 Pourhassan S, Grotemeyer D, Fokou M, *et al*. Extracranial carotid arteries aneurysms in children: single-center experiences in 4 patients and review of the literature. *J Pediatr Surg* 2007;42:1961–8.

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