

# Adams-Oliver syndrome associated with gastrointestinal malformations

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## DESCRIPTION

A male infant presented with drooling of saliva at birth. There was an antenatal history of polyhydramnios. Oesophageal atresia and tracheoesophageal fistula (OA/TOF) were confirmed by the coiled nasogastric tube on chest X-ray. Skin was persistently marbled, regardless of temperature, in keeping with cutis marmorata telangiectatica congenita (CMTC). The second to fifth toes were absent bilaterally; the left great toe hypoplastic (figure 1). Dilated tortuous veins and areas of absent skin,

aplasia cutis congenita (ACC), were evident on the scalp (figure 2). Atrial septal defect was identified on echocardiogram and tortuous retinal vascular pattern on ophthalmology review. Parents were non-consanguineous, but father also had oligodactyly. VACTERL association was considered; however, the characteristic findings led to a diagnosis of Adams-Oliver syndrome (AOS). Surgical repair of OA/TOF was performed on day 2. Contrast study for feed intolerance demonstrated duodenal stenosis requiring duodenoduodenostomy. AOS is rare, with an incidence of 1 in 225 000.<sup>1</sup> ACC and terminal limb defects are typical. CMTC and cardiac malformations are common (20%), while ophthalmological abnormalities occur in <10%.<sup>2</sup> Neurological deficits are uncommon, but more severe vascular phenotypes may have developmental problems and seizures.<sup>3</sup> Different inheritance patterns are described; autosomal dominant likely in this case in view of paternal history. DNA analysis of the six known AOS genes (found in 50% AOS cases) was negative. Physical findings may vary in families due to variable expressivity. This is the first published case of AOS occurring with OA/TOF and duodenal stenosis which may be incidental or an as yet unrecognised association.



**Figure 1** Cutis marmorata telangiectatica congenita of lower limbs with oligodactyly of both feet.



**Figure 2** Multiple areas of aplasia cutis congenita and dilated scalp veins.

## Learning points

- ▶ Thorough systemic evaluation of infants with oesophageal atresia and tracheoesophageal fistula is important to exclude other abnormalities such as VACTERL association (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies and limb abnormalities), CHARGE, Down's and Edward's syndrome.
- ▶ Our patients are not always 'textbook' and there may be other clinical findings and associated conditions. Limb anomalies and cutis aplasia may occur together in Adams-Oliver syndrome (AOS), Goltz and Scalp-Ear-Nipple syndrome.
- ▶ AOS is a rare disorder classically characterised by aplasia cutis congenita and terminal limb defects. It is a disorder that can affect many systems and as such requires multidisciplinary approach to investigation and management.

**Contributors** LvG performed the initial literature search, obtained parental consent and drafted the first manuscript. CG contributed to the literature search and first manuscript. SB arranged medical



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photography and contributed towards management from the surgical team. DD was the senior clinician overseeing the care of the patient. All authors contributed to the editing and review of final manuscript.

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