QUIZ SECTION

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A Child with a Congenital Aplasia of the Scalp: A Quiz

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A 3-year-old boy was referred to paediatric dermatology for a congenital aplasia of the scalp. He had no family history and was born at term. He was the second child of non-consanguineous parents. Physical examination showed aplasia cutis congenita of the scalp (Fig. 1A, B). It also revealed reticulated, well-demarcated red-purple plaques, with a coarse fixed livedo pattern that was consistent with cutis marmorata telangiectatica congenita of the right arm, trunk, and legs (predominantly on the right leg) (Fig. 1C). Brachydactyly with short distal phalanges and mild cutaneous syndactyly was present (Fig. 1D). Neurological evaluation revealed global developmental delay (acquisition of walking at 22 months, disability in fine motor skills, no word association at age 3 years) and epilepsy. Findings were normal for the remaining clinical examination. Brain MRI showed right aplasia cutis with a defect of the bone (Fig. 1E), haemorrhagic lesions of the choroid plexus, and cortico-subcortical atrophy. Results of echocardiography and abdominal ultrasonography were normal. Ophthalmology examination did not reveal any retinal anomalies.

What is your diagnosis?

Differential diagnosis 1: Adams–Oliver syndrome

Differential diagnosis 2: Amniotic band sequence

Differential diagnosis 3: Focal dermal hypoplasia

Differential diagnosis 4: Macrocephaly cutis marmorata telangiectatica congenita

See next page for answer.

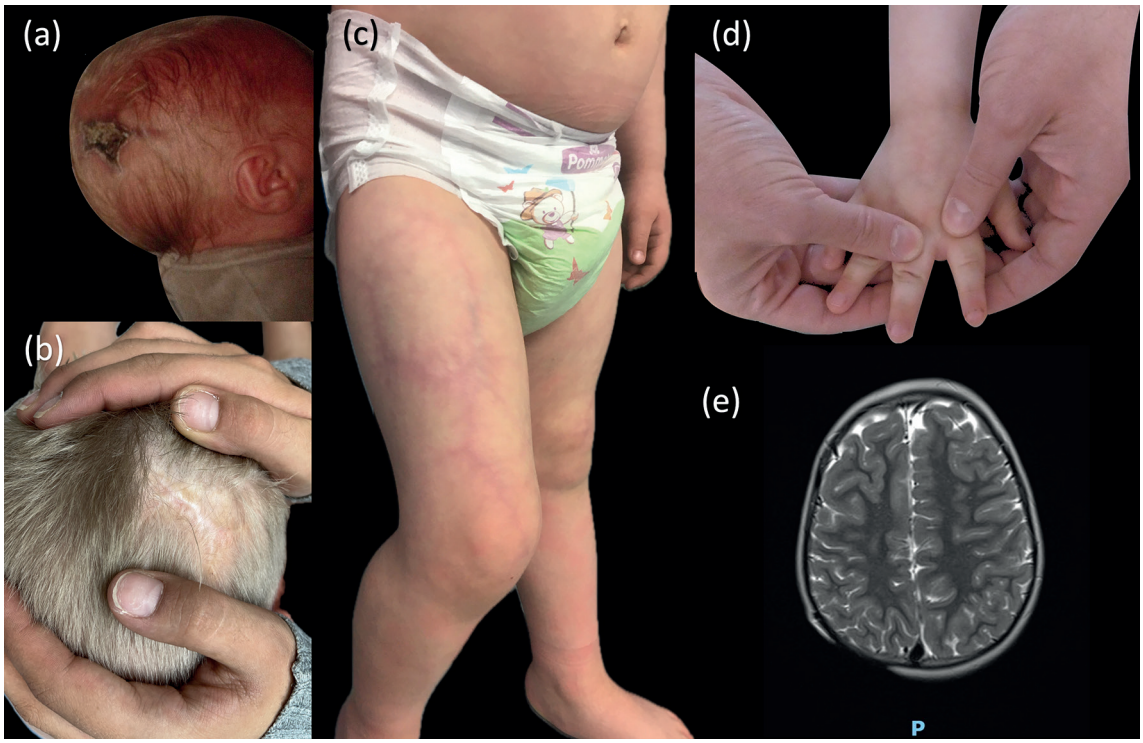

Fig. 1. (a) Scalp defect at birth (aplasia cutis congenita). (b) Aplasia cutis congenita. (c) Cutis marmorata telangiectatica of the right leg. (d) Shortening of terminal phalanges of fingers (brachydactyly). (e) Brain MRI (axial section, T2-weighted image) showing a defect of the parietal bone.

ANSWERS TO QUIZ

A Child with a Congenital Aplasia of the Scalp: A Commentary

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Diagnosis: **Adams–Oliver syndrome**

Adams–Oliver syndrome (AOS) (OMIM PS100300) is a rare congenital condition characterized by the association of congenital scalp defects (aplasia cutis congenita) and terminal transverse limb defects (1). The estimated incidence is 1 per 250,000 live births (2). Other anomalies associated with AOS include cutis marmorata telangiectatica congenita (3), dilated tortuous veins on the scalp (3), congenital heart disease (ventricular and atrial septal defects, coarctation of the aorta, bicuspid aorta, etc.) (4), ocular anomalies (retinal detachment, microphthalmia, visual impairment, optic atrophy) (5), and neurological involvement (cognitive disability, spastic hemiplegia or diplegia, seizure) (6). Several pathogenic variants with autosomal dominants forms have been identified (NOTCH1, DLL4, ARHGAP31, RBPJ) as well as autosomal recessive forms (DOCK6, EOGT) (7).

The clinical severity of AOS is heterogeneous, with inter- and intra-familial phenotype variability. Ocular and neurologic anomalies are frequent in patients with DOCK6 variants and heart disease in patients with NOTCH1, DLL4, and RBPJ variants (7). However, only 30% to 50% of patients with AOS have a pathogenic variant (7), and *de novo* pathogenic variants have been reported, but the frequency is unknown (1). In our patient, genetic analysis did not reveal any pathogenic variant in the genes currently identified in this pathology.

Management requires a multidisciplinary approach (paediatric neurologists, paediatric cardiologists, ophthalmologists, dermatologists, plastic surgeons) for investigation and management. A complete imaging assessment (echocardiography, abdominal ultrasonography, brain MRI) should be performed to detect abnormalities and provide early care (1). In addition, genetic counselling should be offered to families affected by this disease.

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