Aplasia cutis: clinical, dermoscopic findings and management in 45 children

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Aplasia cutis: clinical, dermoscopic findings and management in 45 children

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Editor,

Aplasia cutis congenita(ACC) is a rare malformation characterized by a localized absence of skin. This condition, most frequently seen on the scalp, shows a variety of clinical features, from fissure-like ulcers to atrophic macules, and can be solitary or multiple. Specifically, a cystic variant covered with a translucent thin and round surface has been named membranous aplasia cutis (MAC) by Drolet et al[1].

In fact, MAC may present distinctive clinical signs related to cranial dysraphism and split cord malformation: hair collar sign, tuft sign and vascular stain[1,2]. On the other hand, ACC presenting with large, irregular or stellate scarring defect has been called non-membranous or classic aplasia cutis, often associated with defects of the underlying bone, with exposure of the dura, sagittal sinus, and brain.

The aim of this case series is to provide clinical and dermoscopic findings of ACC in order to promptly identify this condition and to perform diagnostic investigations.

Medical records and photographs were collected in this single center retrospective study of patients in the Pediatric Dermatology Unit of the University of Bologna (Italy), from 2004 to 2019.

Forty-five children affected by ACC were recorded (Table 1). In our sample one case of familial AAC was found (Figure 1a).Seven patients presented more than one lesion: 5 children with 3 lesions, 2 children with 2 lesions for a total of 57 ACCs (Figure 1b). Within our sample, 96.5% of ACCs were localized on the scalp. Seventeen cases of classic ACC presented as scarring alopecia

oval (4/17) or star-shaper (13/17) ; 2/17 cases of scarring ACC involved a large area of the scalp (>5cm) and required a neurosurgical approach.Forty cases of non-scarring ACC were detected and appeared oval in shape with a diameter ranging from 0.7 to 3.2 cm (Figure 1c,e,g). Hair collar sign and nevus flammeus vascular stain with a small red nodule were detected alone or concomitantly with non-scarring ACC (Figure 1d).

Unlike classic ACC dermoscopy, which shows an area of scarring alopecia, the non-scarring ACC dermoscopy is more challenging. In our case series we have detected the known membranous pattern of ACC in 14 cases (Figure 1f) but also a new "pseudomembranous" pattern in 24 children (Figure 1h).

Ultrasonography, skull X-ray and brain MRI were respectively used in 24, 12 and 11 children. In most children the defect was exclusively cutaneous, whereas in 2/45 there was also an involvement of the underlying bone. In 3 children with non-scarring ACC associated with hair collar sign plus nevus flammeus we respectively detected: (i) a cranial meningocele, (ii) malformation of the venous system of the occipital area (iii)syndactyly and brachydactyly (Adam-Oliver syndrome).

In conclusion, 91.11% of children presented a scalp ACC without multiple anomalies (Type 1 according to Frieden et al)[3].

Scarring and non-scarring ACCs of the scalp are often sporadic conditions, but familial occurrences have been documented[4].Adams–Oliver syndrome is characterized by wide spectrum of ACC, especially scarring ACC, and terminal transverse limb defects[5,6].

This study highlights the value of dermoscopy in the detection of ACC and in the differential diagnosis with other forms of localized hair patches[7,8]. In our experience, we identified a dermoscopic aspect that we named pseudomembranous pattern, which can be included within a clinical and dermoscopic spectrum ranging from the classic to the pure membranous form of ACC. However, this new pattern was not associated with an increased risk of malformations.

In the diagnostic algorithm of ACC, an accurate dermatological evaluation of all the body represented the first step. In addition, ultrasound could be useful for the diagnosis of mild ACC and for the screening of ACC-associated skull bone defects[9]. However, in patients with ACC associated with hair collar sign or vascular stain with a small nodule, especially when more than one cutaneous sign occurs simultaneously or if ultrasound is positive or suspicious for occult cranial dysprafism, brain MRI is mandatory[10].

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The patients in this manuscript have given written informed consent to the publication of their case details.

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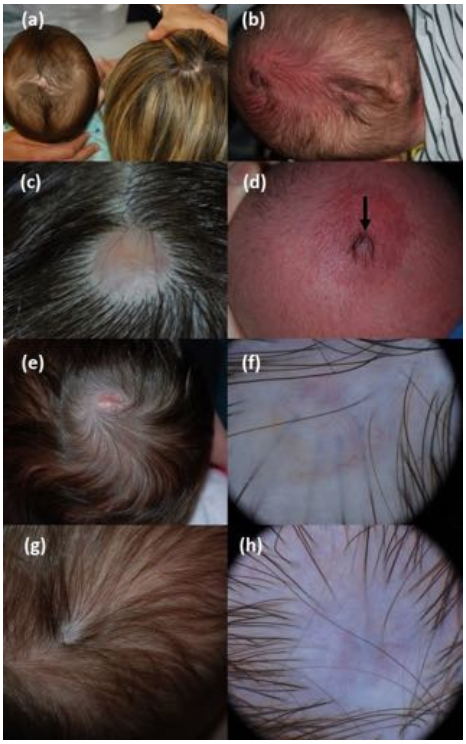
Figure 1: (**a)**Non-scarring aplasia cutis on the vertex of a mother with her son without association with multiple abnormalities;**(b)** a child with three concomitant non-scarring ACC localized on the vertex and occipital bone associated with hair collar sign and nevus flammeus; **(c, e, g)**three cases of non-scarring, oval shaped ACC localized on the vertex **(d)**non-scarring ACC associated with hair collar sign and vascular stain with a small red nodule on the upper part; **(f)**dermoscopy shows a roundish patch of alopecia with some telangiectatic vessels at the centre.At the periphery, radially and horizontally oriented hair follicles (starburst pattern) were clearly visible from the bulbs to the follicular ostia, forming the membranous pattern of ACC; **(h)**dermoscopyhighlights the pseudomembranous pattern characterized by unfocused teleangectasic vessels at the centre and radially oriented hair follicles without evidence of the bulbs.

Table 1: Epidemiological, clinical and dermoscopic findings of aplasia cutis in 45 children

Characteristics of patients	**Lesions of ACC**	**Non-scarring ACC**		**Scarring (classic) ACC**
		Membranous	**Pseudomembranous**	
	57	14	26	17
EpidemiologicalFindings				
Age (months), mean ± SD	12,35 ± 25,06	13,3 ± 20,03	12,5 ± 4,3	7,2 ± 8,9
Male gender, n (%)	22	4	6	12
Anatomical site				
Scalp	55	13	25	17
vertex	25	8	10	7
Left parietal bone	13	3	6	4
Right parietal bone	10	2	5	3
Occipital bone	5	2	2	1
Frontal bone	2	1	1	0
Back	2	0	0	2
Associated clinicalsigns				
Hair collar sign	9	4	5	0
Fauntail	0	0	0	0
Nevus flammeus + Hair collar sign	7	3	4	0
Little nodule + Nevus flammeus + Hair collar sign	2	1	0	0

Dermoscopic features	45	16	24	5
Starbust pattern	42	16	24	0
Telangectatic vessels	26	16	10	0
Follicular bulbs clearly visible	16	16	0	0



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