Pediatric Dermatology - Interactive Case Workshop

European Dermatology Forum
EURODERM excellence

Euroderm Excellence, November 2025, Rome

Dario Francesco D'Urso (Rome) & Dagmar Jamiolkowski (Hannover)

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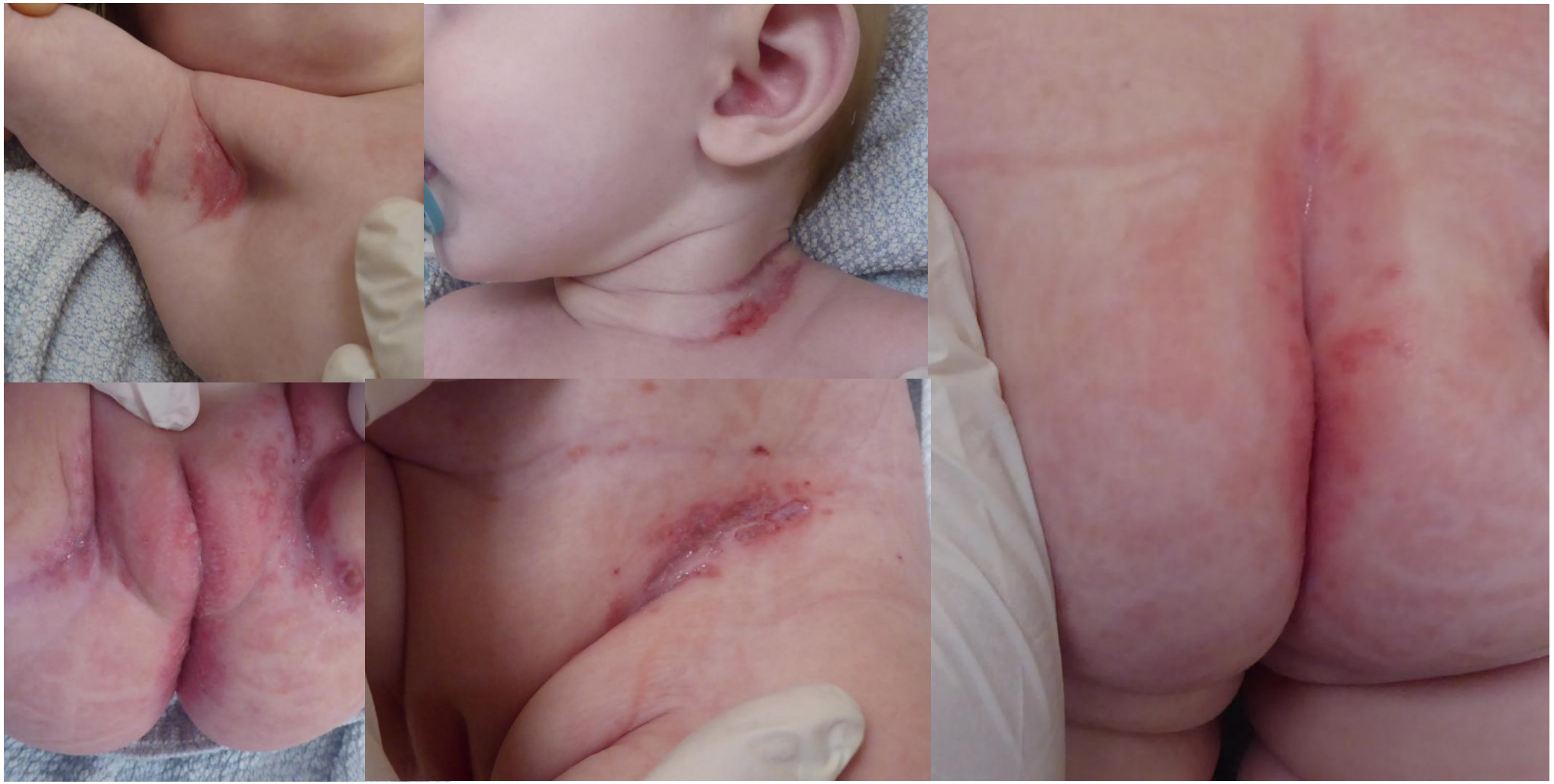

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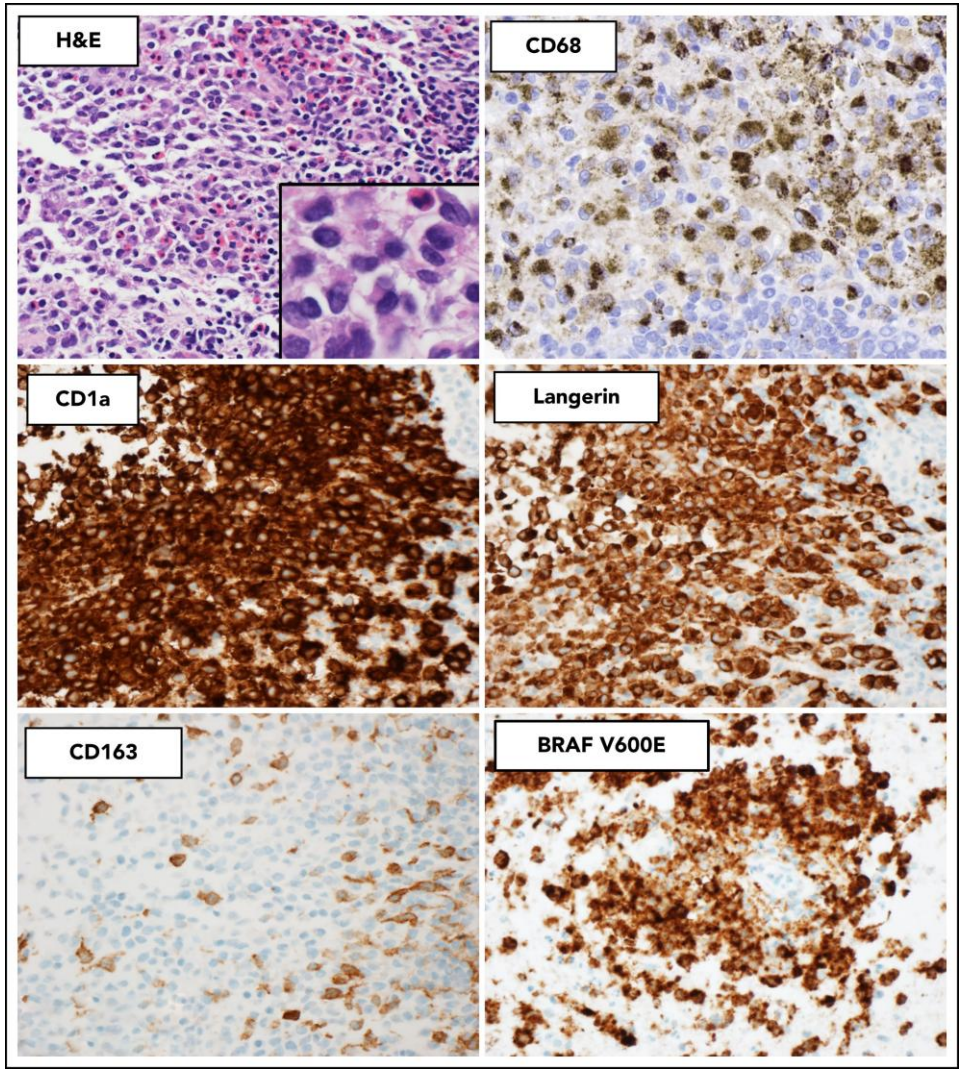
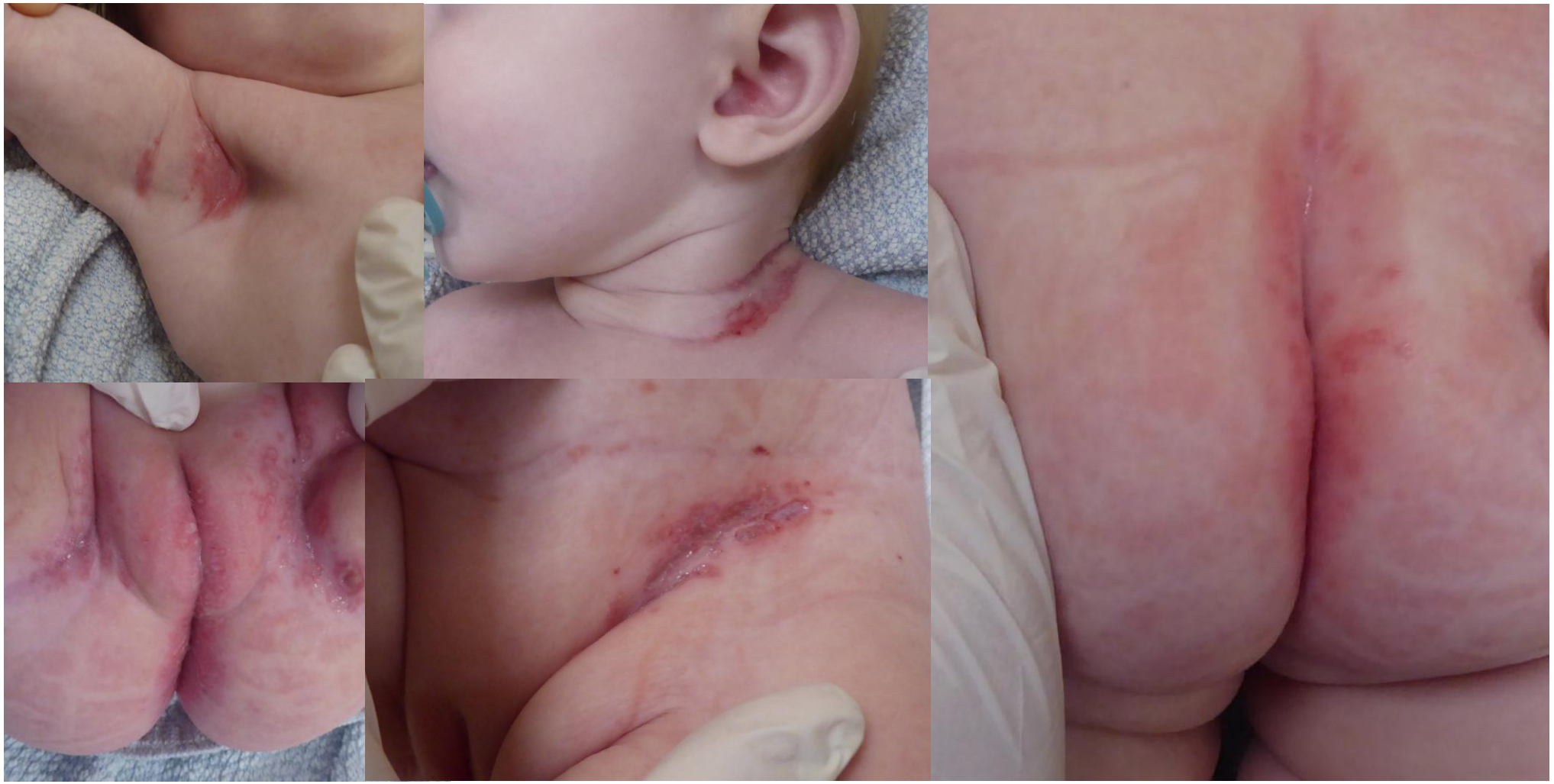


- Lesional skin biopsy:
  - CD1a, S100, Langerin, BRAF V600E/E2/D
- Initial whole-body MRI:
  - normal
- Whole-body MRI after 3 months:
  - bone lesion in scapula and hepatic lesion

Goyal G *et al. Blood* 2022

Multisystem Langerhans cell histiocytosis (MS-LCH)



Multisystem Langerhans cell histiocytosis (MS-LCH)

Type	# of organs involved	# of lesions per organ	
Single-system unifocal	1	1	
Single-system multifocal	1	≥2	
Single-system pulmonary	Lungs	≥1	
Single-system CNS	CNS	≥1	
Multi-system	≥2 Involvement of liver, spleen, and bone marrow = high risk	≥1	

Therapy of skin lesions

- TCS, TCI

Therapy of MS-LCH (ped. oncology protocols):

- prednisolon, vinblastine; 6-mercaptopurine, methotrexate

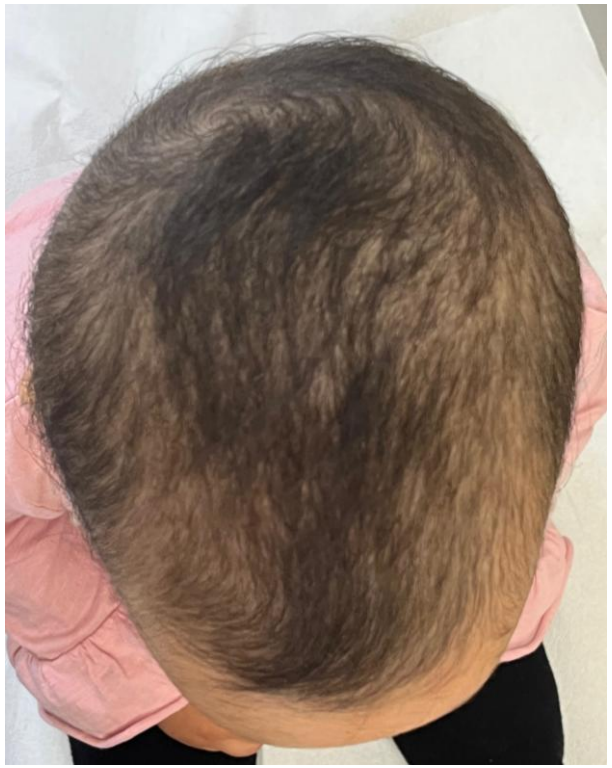
Sconocchia T *et al. Front Immunol.* 2023

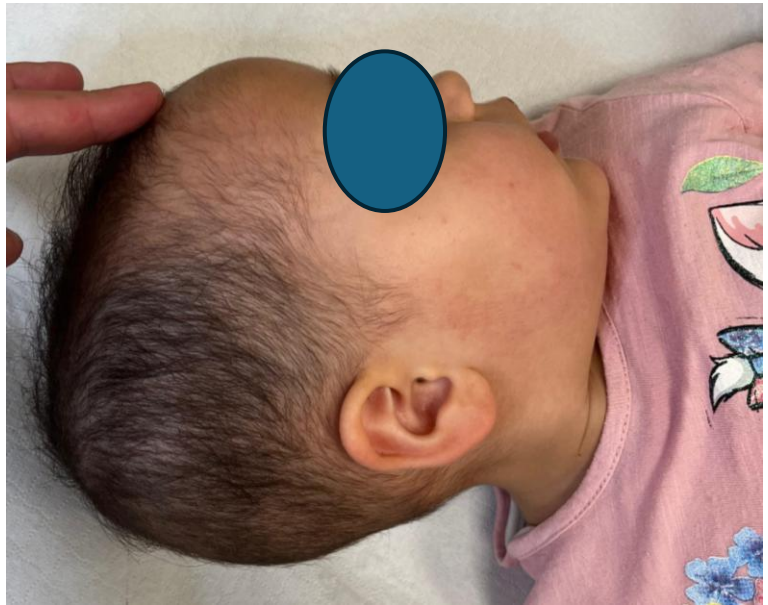
Female patient, 2 years and 2 months old, born at term by cesarean section

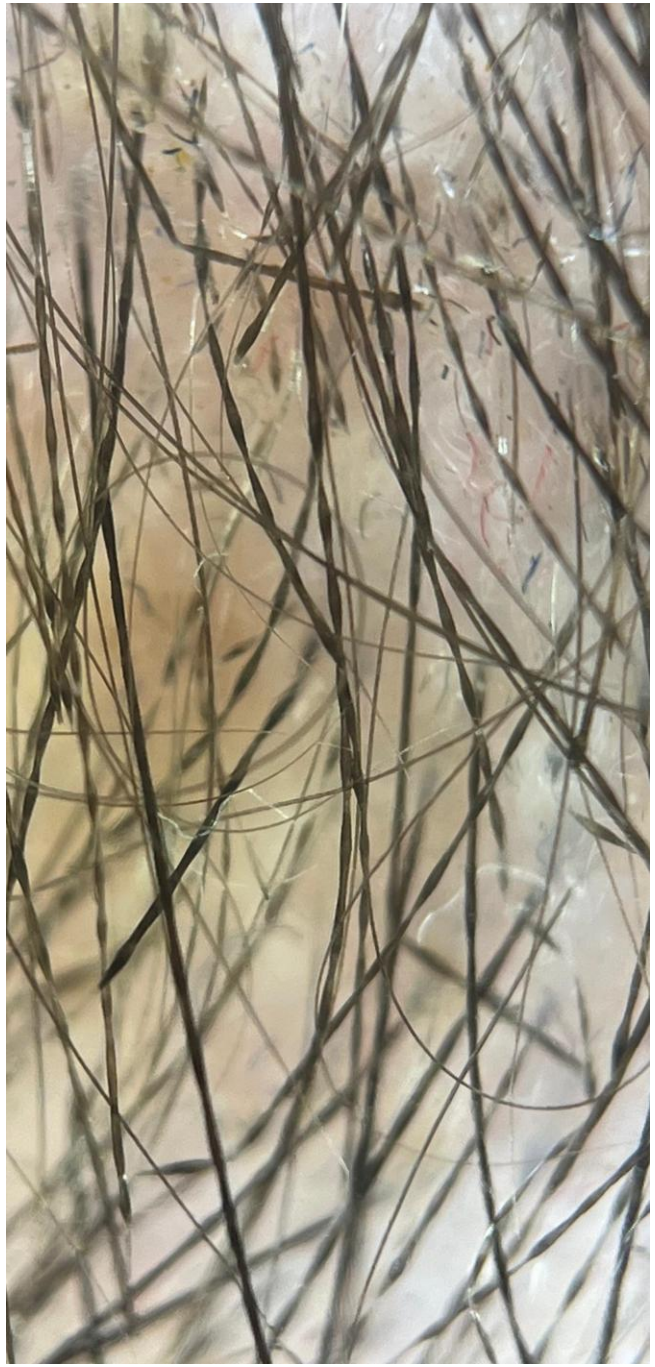
The mother reports poor hair growth for the past year.

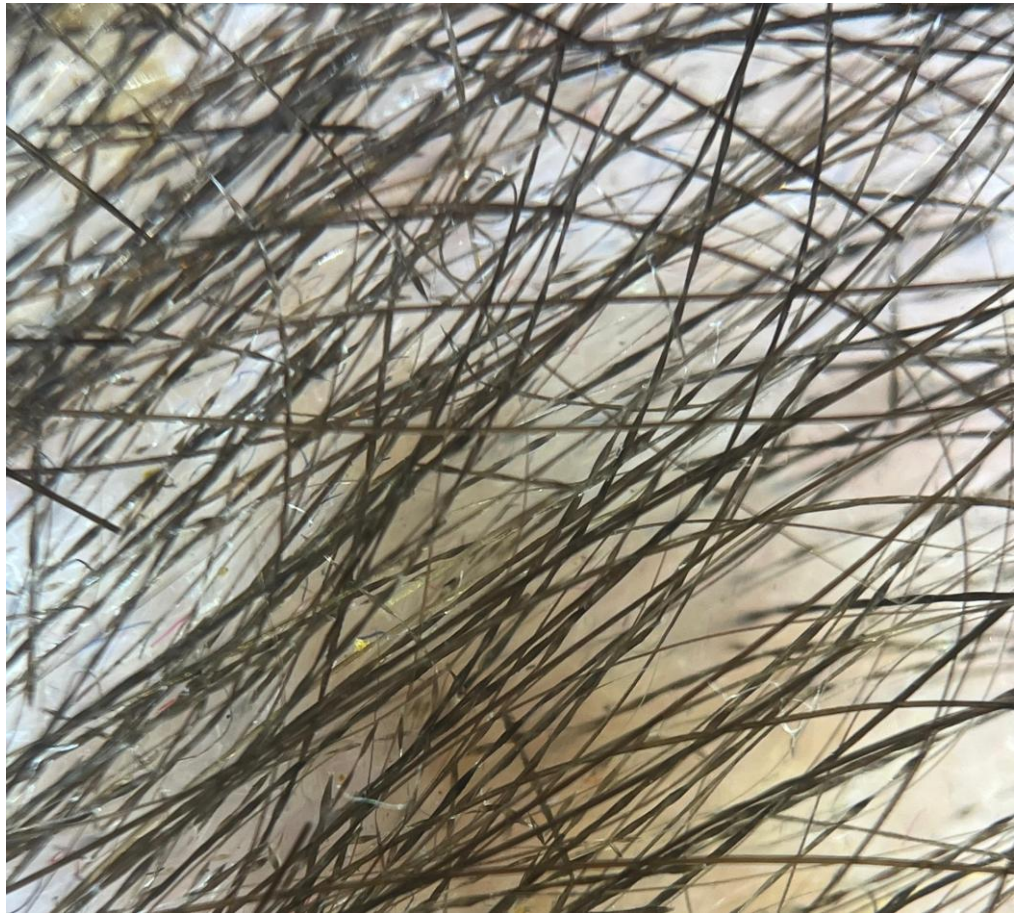
At birth, there was a normal presence of hair

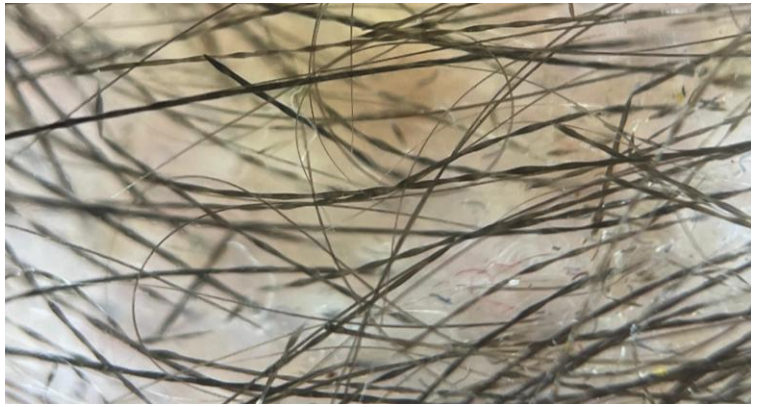
No other abnormalities involving skin or skin appendages.











Diagnosis

1 – Trichorrhexis nodosa

2 – Trichorrhexis invaginata

3 – Trichothiodystrophy

4 – Monilethrix

5 – Carvajal syndrome

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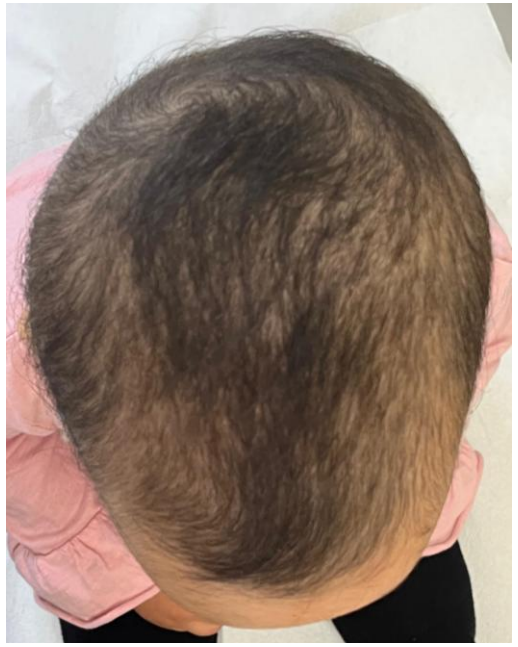
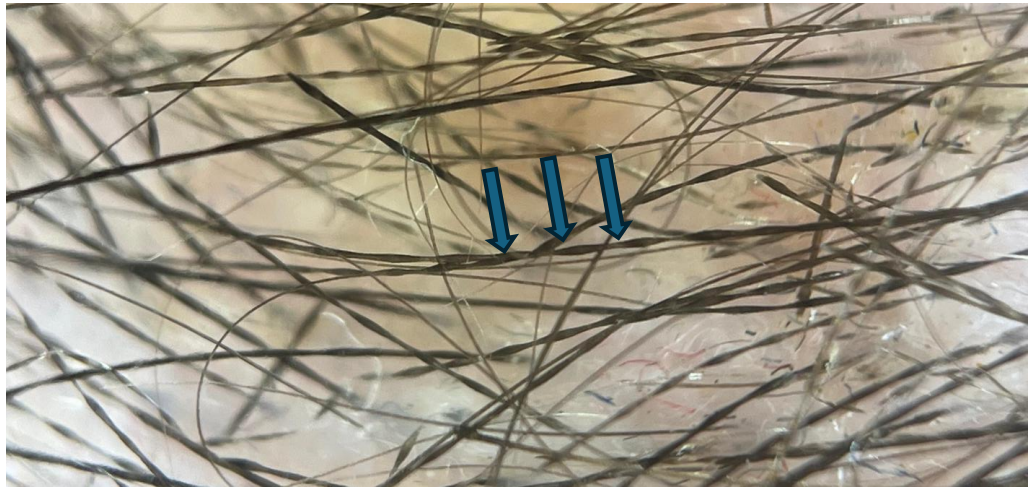


MONILETHRIX

Lock of hair characterized by dystrophic constrictions regularly separated by elliptical nodes of normal thickness, giving it a moniliform (beaded) appearance.

NM_002281.4 (KRT81): c.839C>T (p.Ala280Val) in heterozygosity with paternal segregation;
Pathogenic variants in the KRT81 gene are associated with Monilethrix with autosomal dominant inheritance (OMIM: 158000).



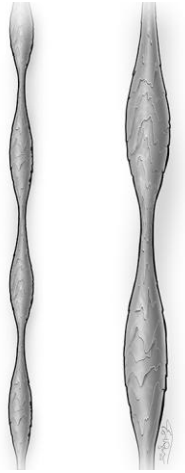
Courtesy of Sabrina Rossi MD

MONILETHRIX

Autosomal dominant mutations in keratin genes (KRT81, KRT83, KRT86) and desmoglein 4 (DSG4).

Impaired formation of intermediate filaments.

Hair is normal at birth (when lanugo hair is replaced by terminal hairs) but becomes short and fragile during the first year of life.



A reduced hair density is observed, particularly in the occipital region. Eyebrows and eyelashes may also be affected.

The prognosis is variable. Sometimes it improves at puberty, but in most cases it persists

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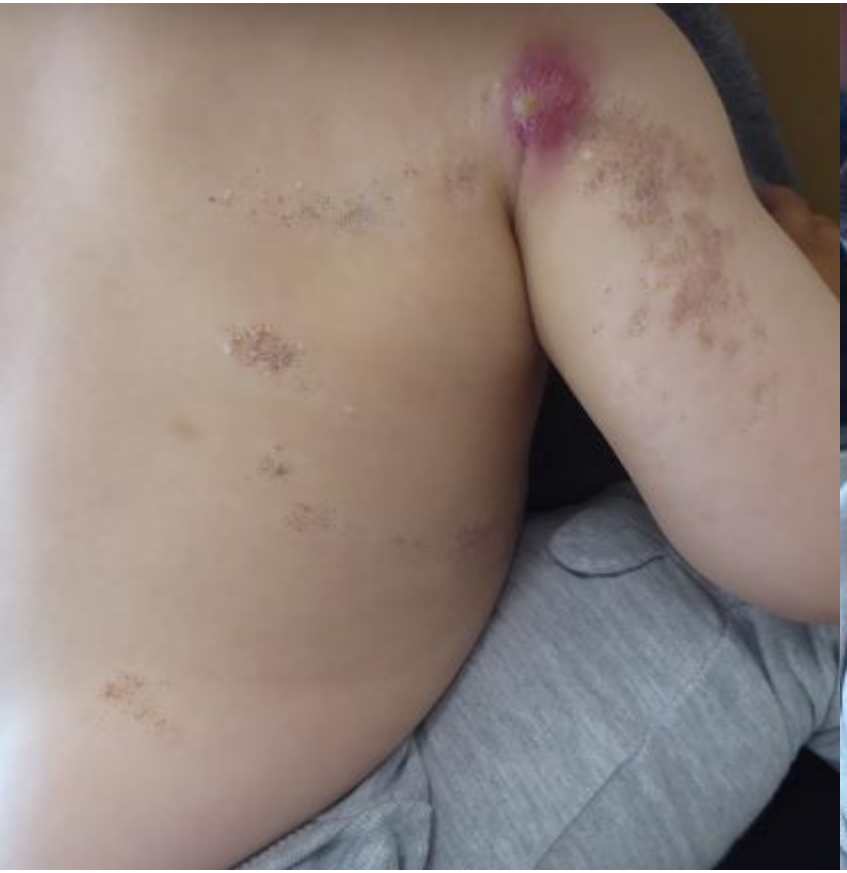
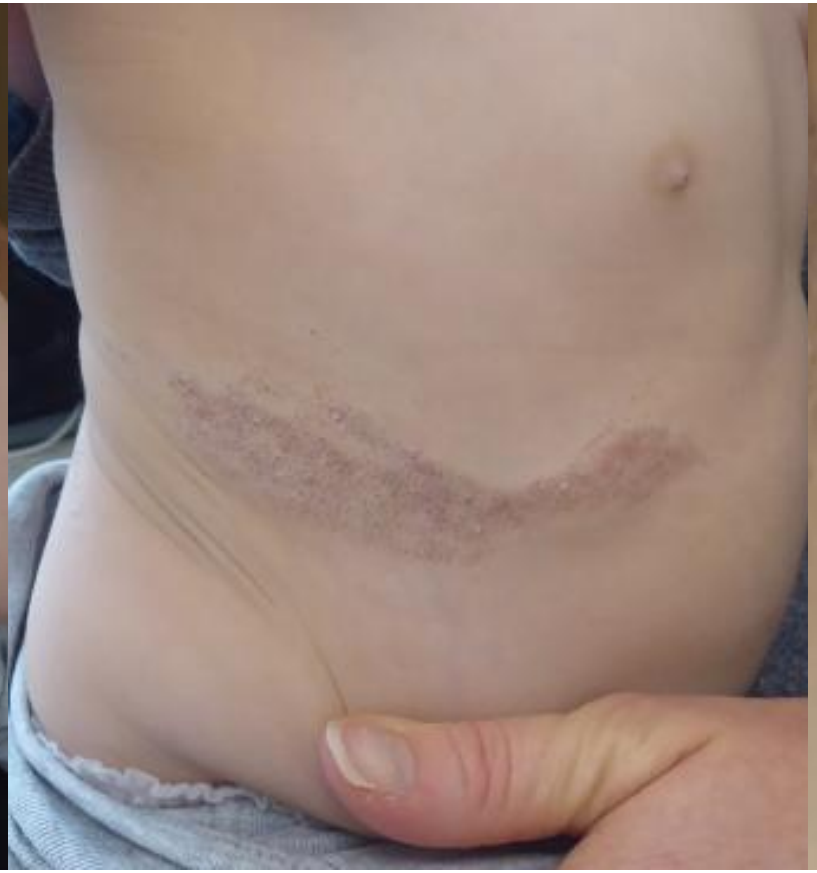
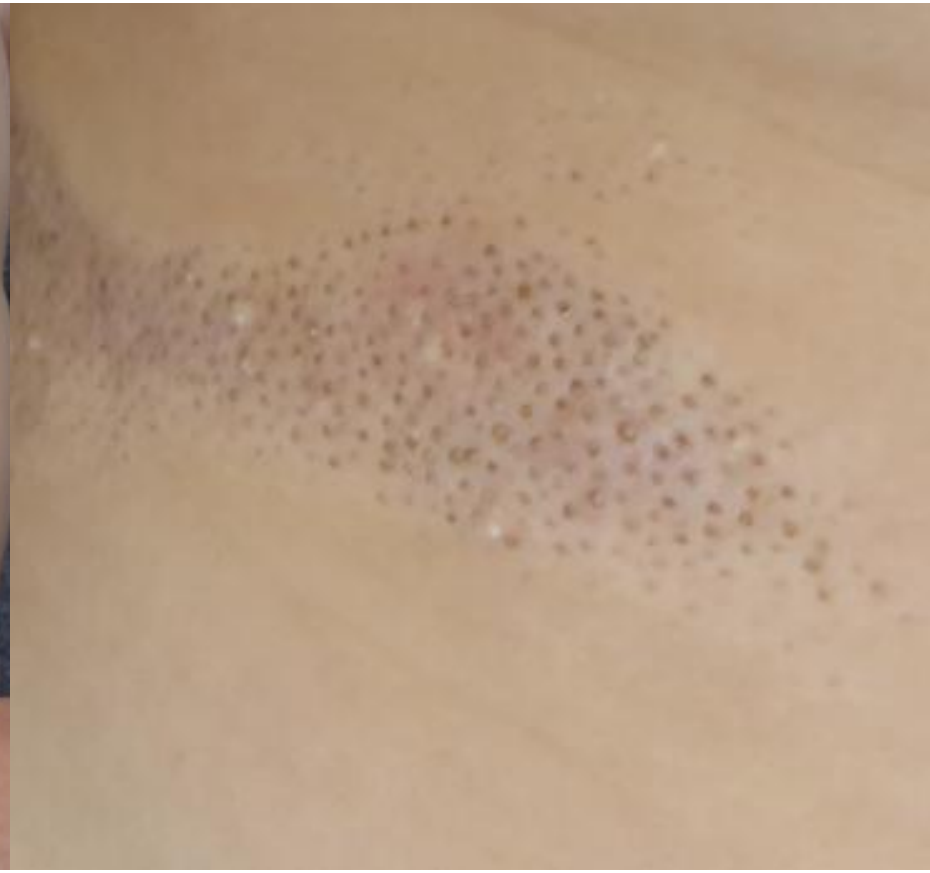
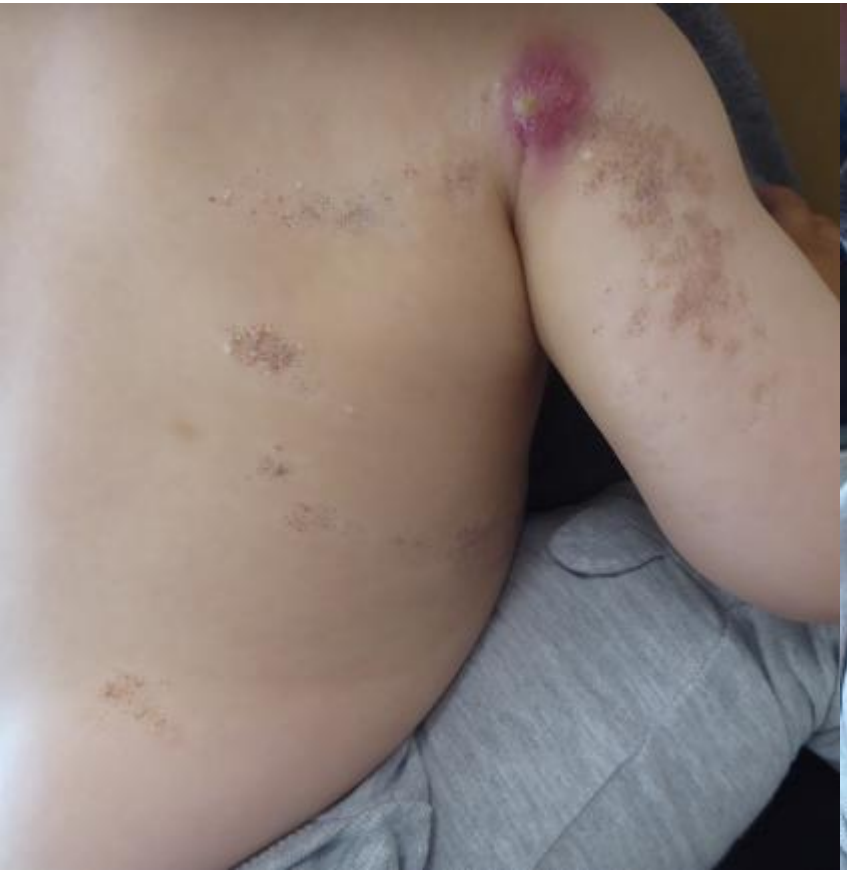
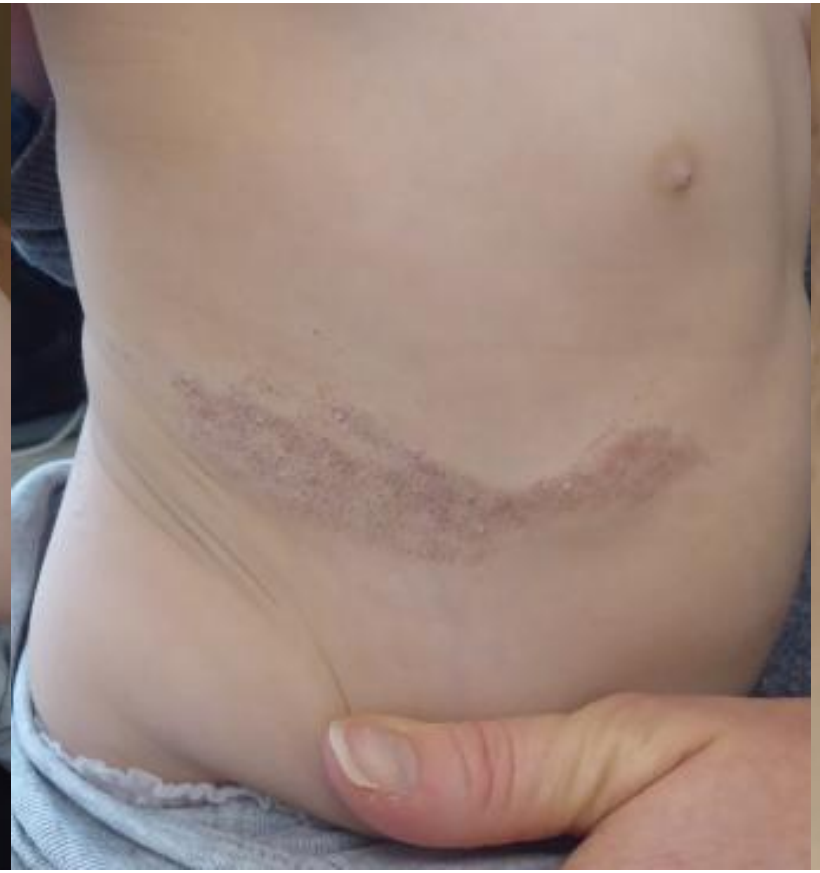
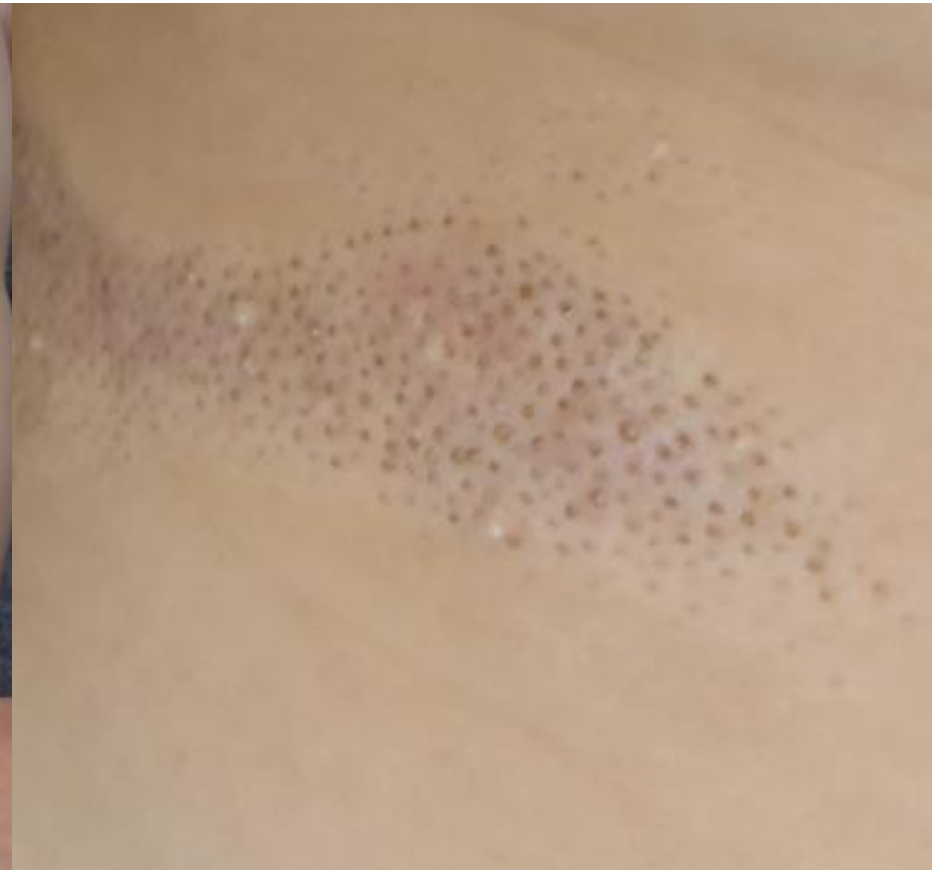




Blaschkolinear nevus comedonicus

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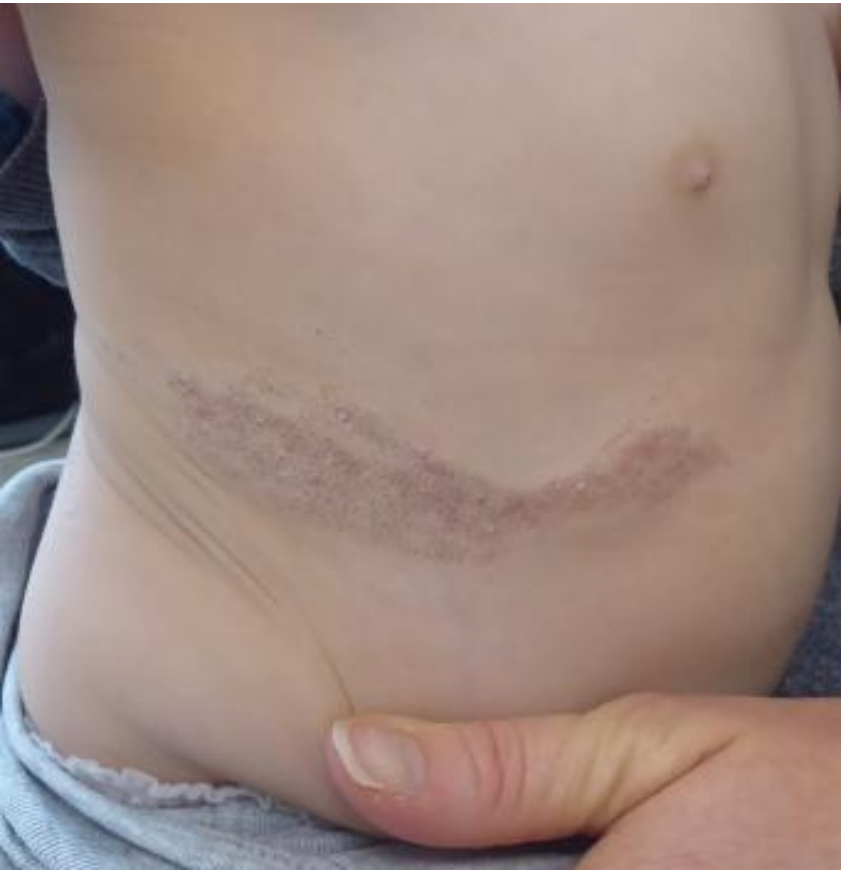
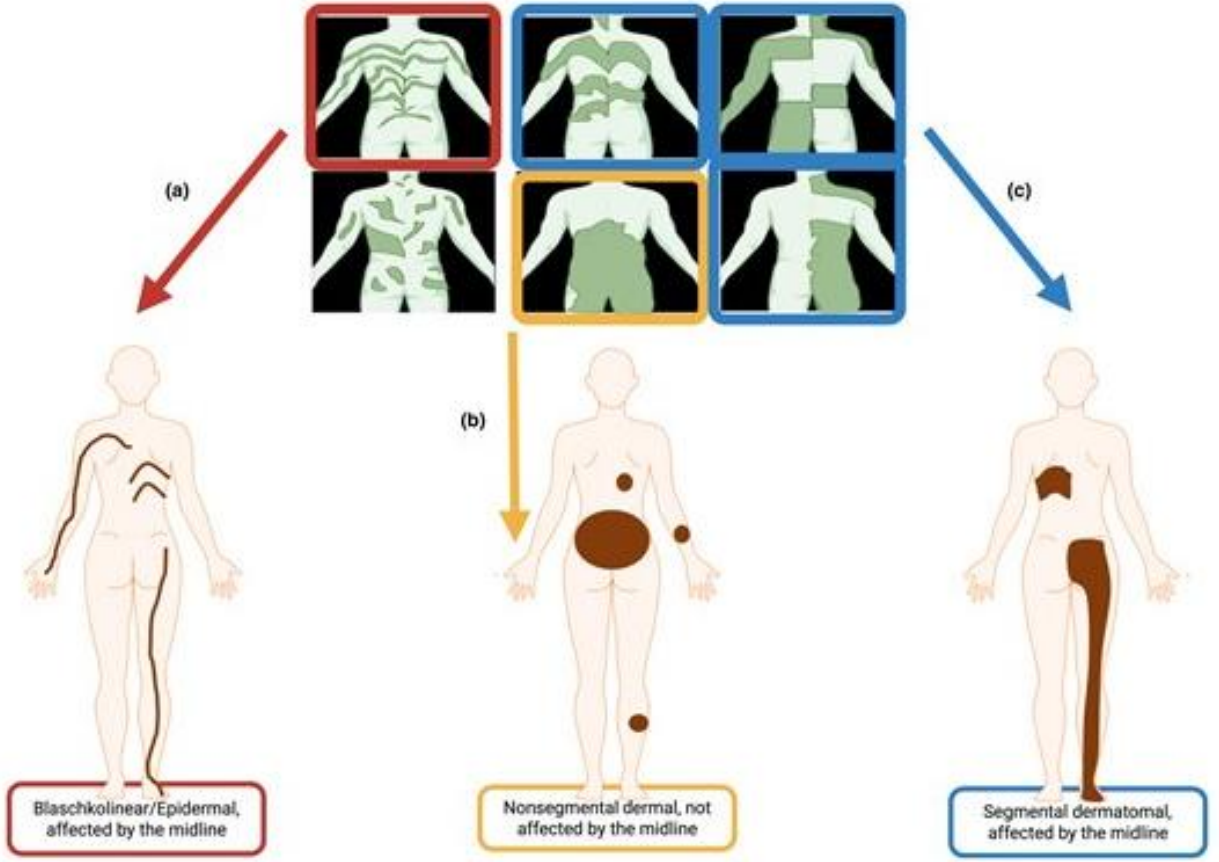
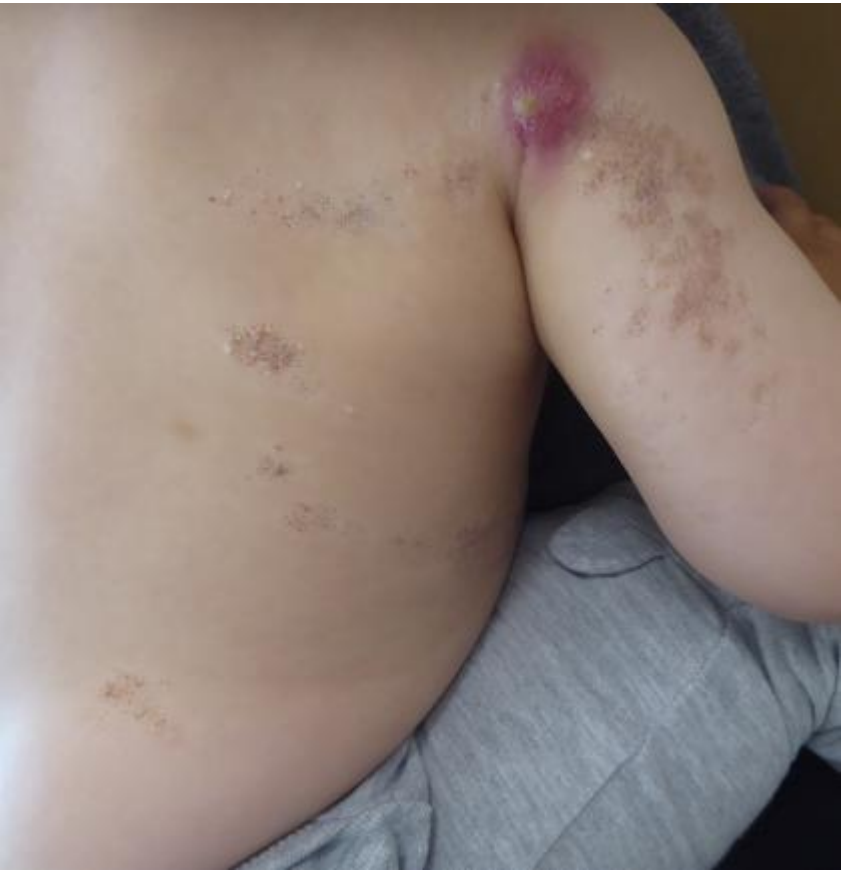




Blaschkolinear nevus comedonicus



Rare epidermal nevus type (mosaic *NEK9)*

Check for

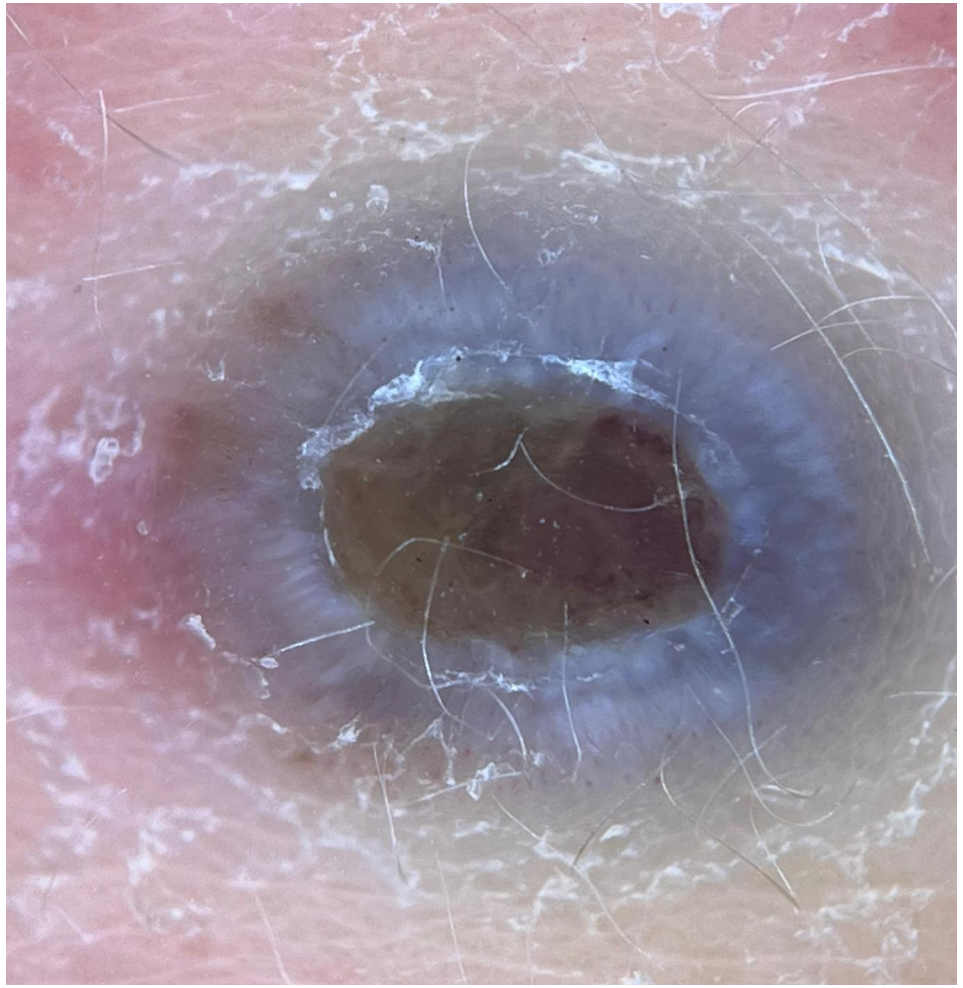
- NC syndrome: ophthalmologic (cataract), neurological (IQ, seizures), skeletal abnormalities (mainly akral)
- Basal cell carcinomas, trichoepithelioma, keratoacanthoma

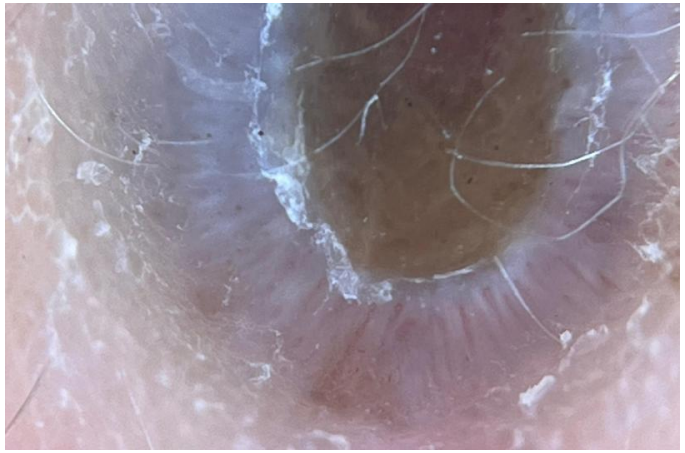
Treatment options

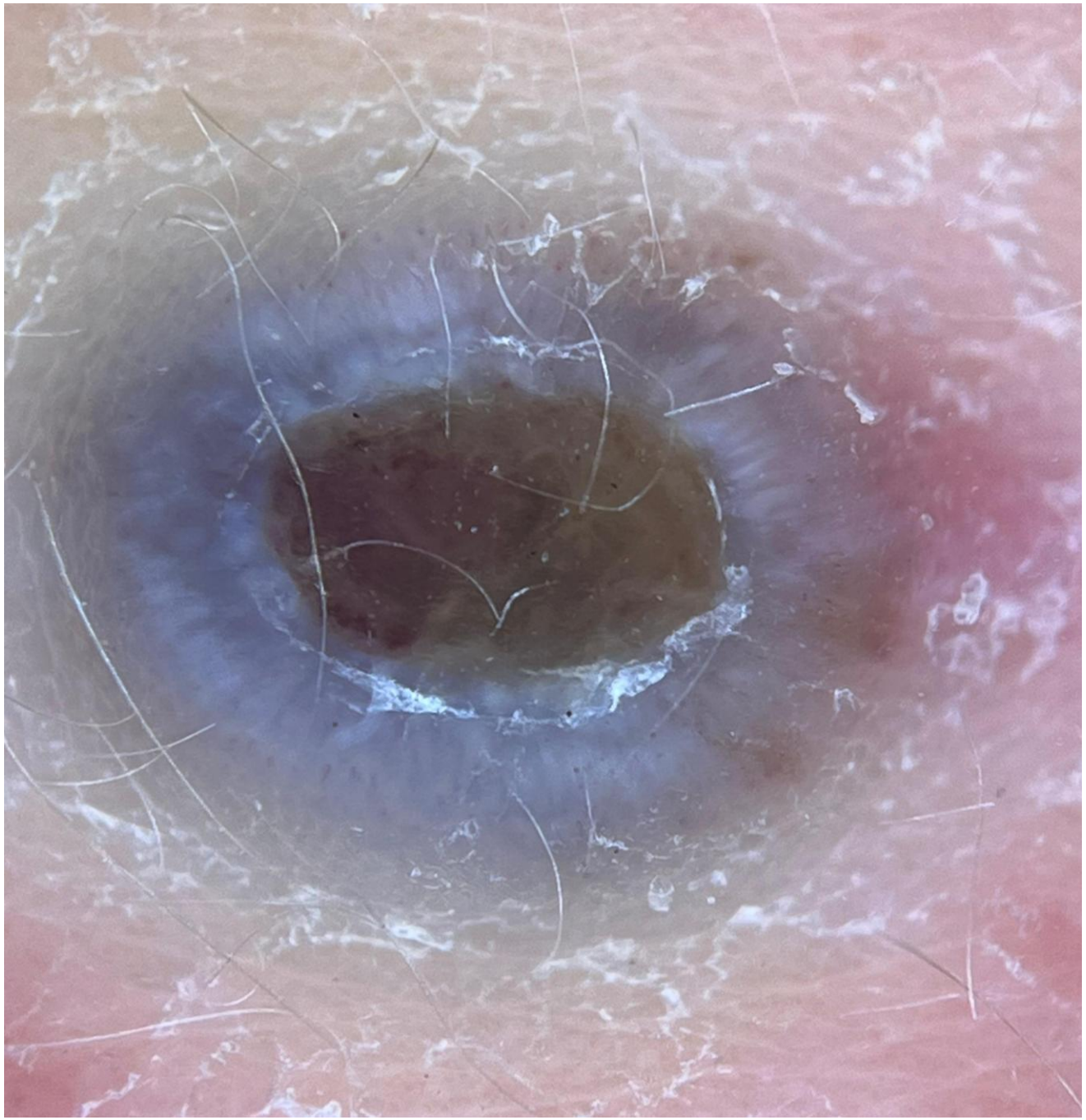
- Topical treatment, laser, surgical approach
- Benzylperoxide 3% gel
- Retinoids not effective

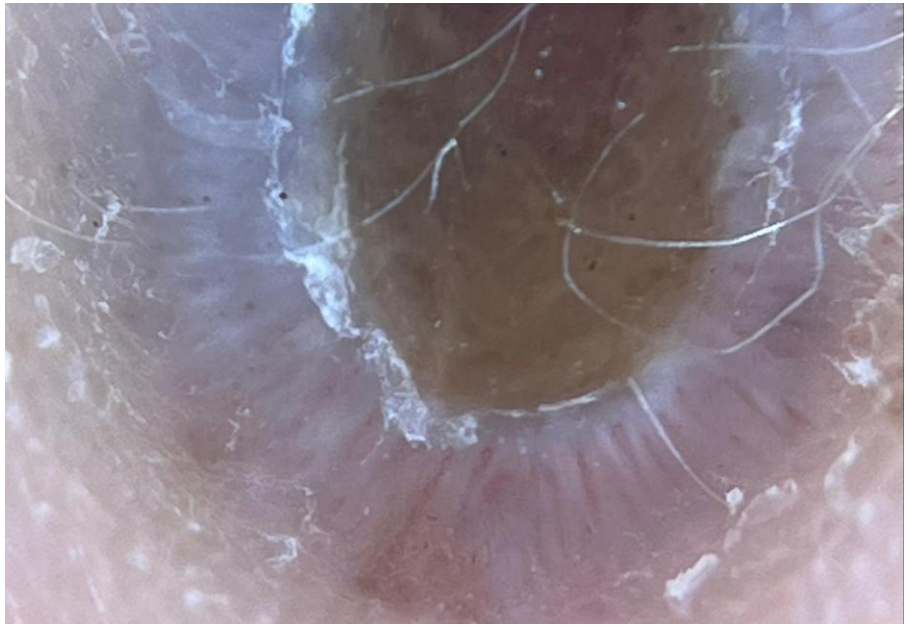
Juratli HA *et al.* J Am Acad Dermatol. 2022; Torchia D. Pediatr Dermatol. 2021

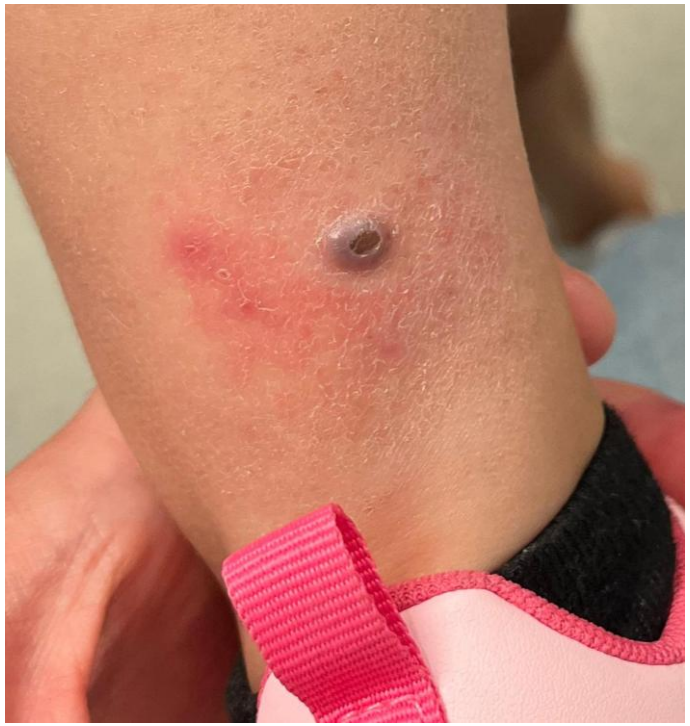
Female patient, 2 years and 2 months old, born at term by cesarean section in good condition.
Two months previously onset of a nodular lesion on the posterior region of the leg.
Previous diagnosis – in another Hospital- of pyogenic granuloma
Treatment with clobetasol propionate











Diagnosis

1 – KAPOSIFORM HEMANGIOENDOTHELIOMA

2 – NEUROTHEKEOMA

3 – TRICOBLASTOMA

4 – MELANOMA

5 – ANEURYSMAL DERMATOFIBROMA

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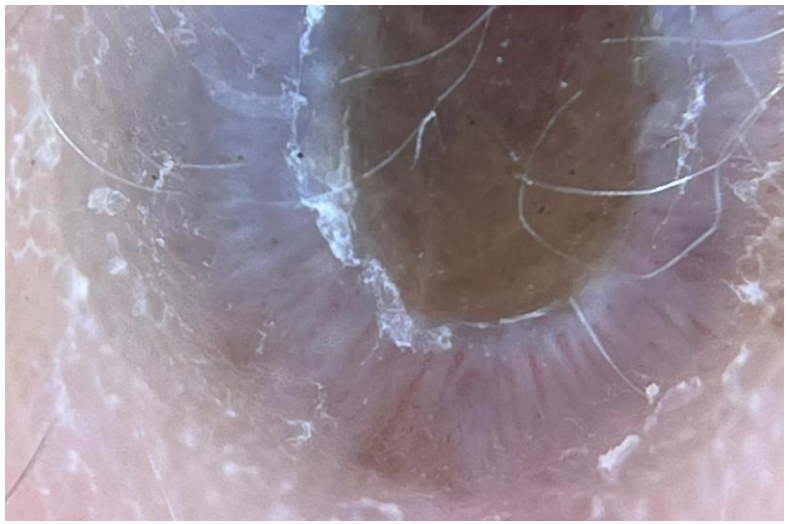
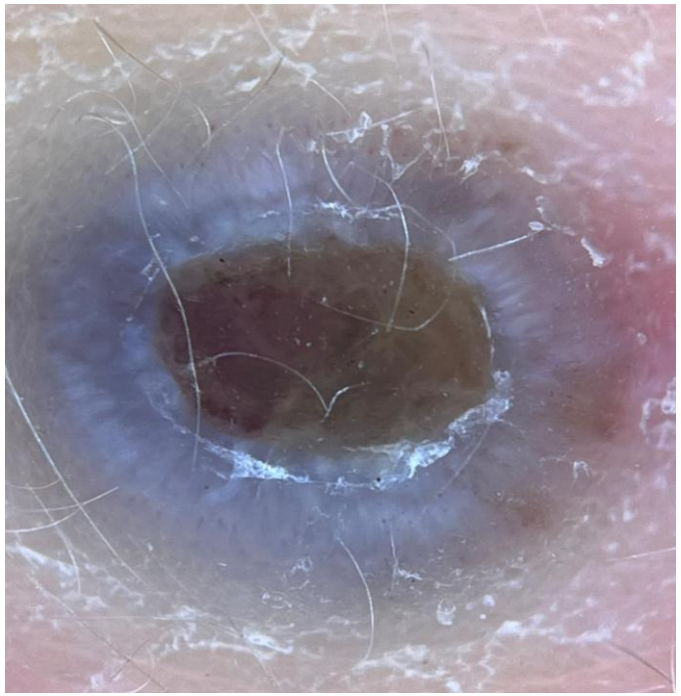
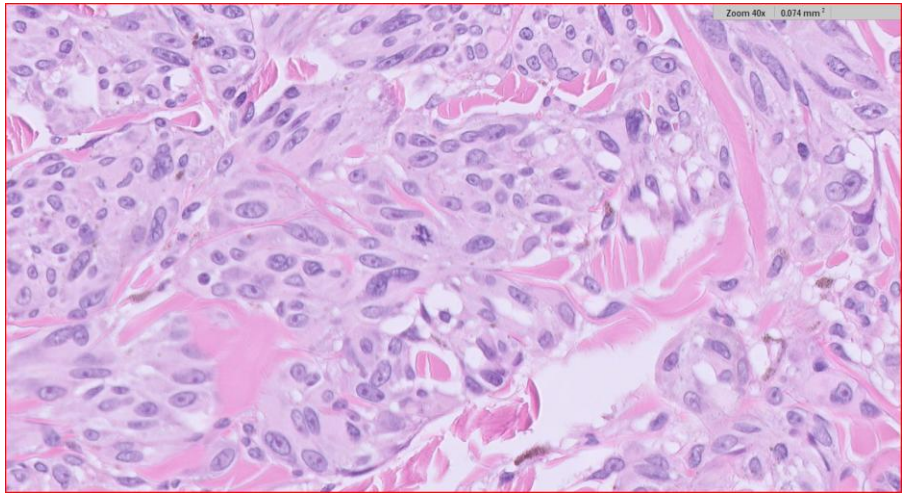
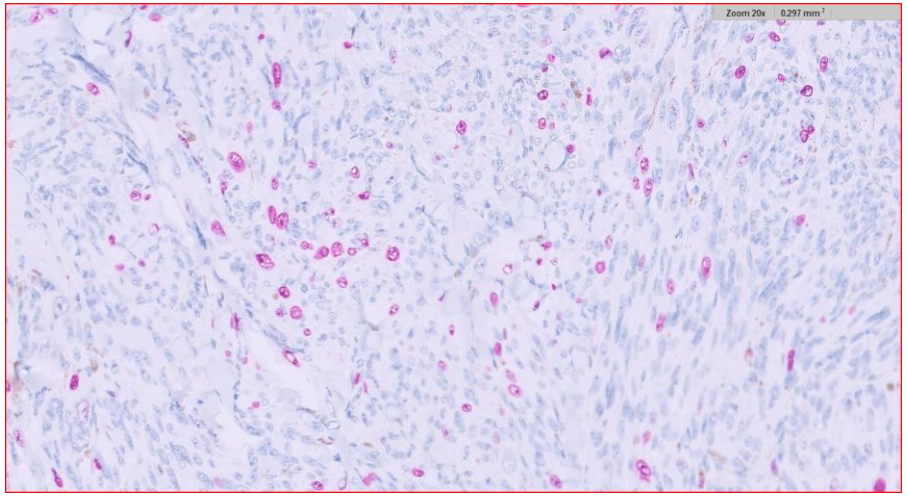
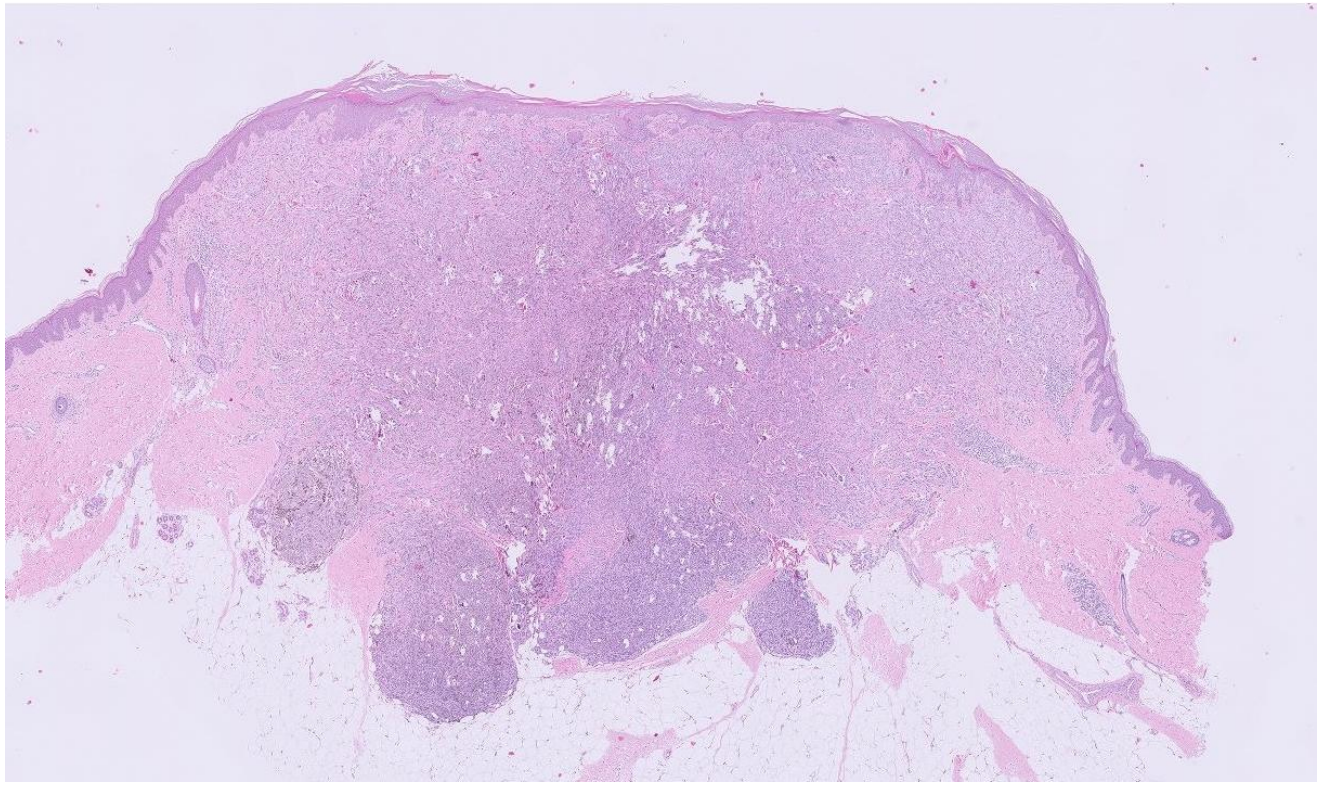








NODULAR MELANOMA
BRESLOW MM 3.8.
PT3A



The neoplasm was investigated with multiple molecular analyses, and the case was re-evaluated within the framework of the MELCAYA study group (Melanoma in Children, Adolescents and Young Adults).

Courtesy of Sabrina Rossi

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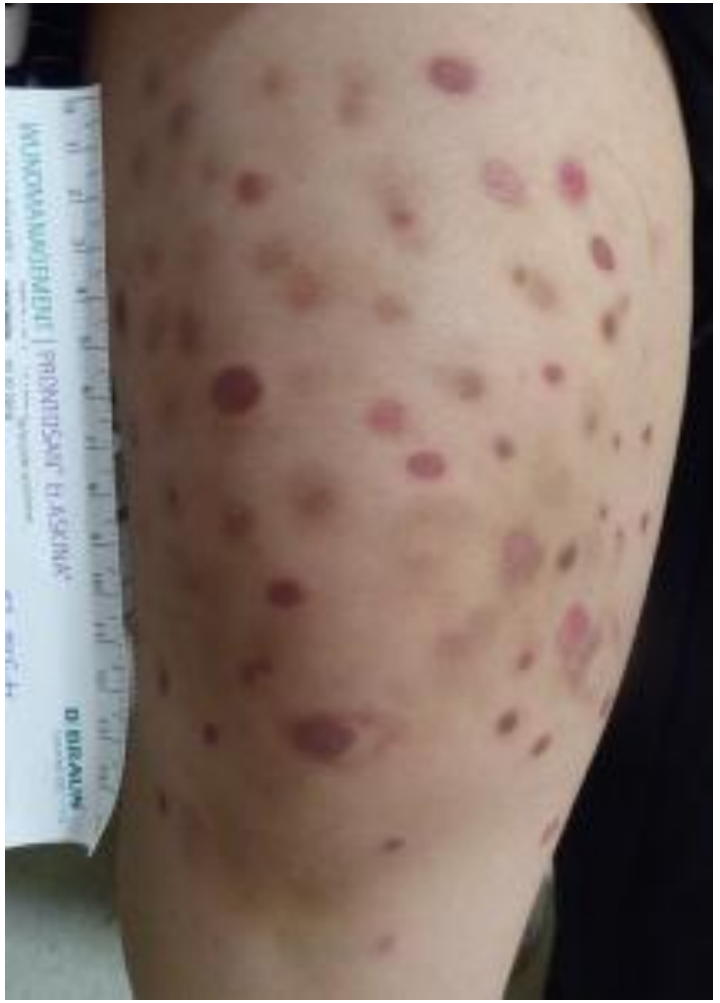

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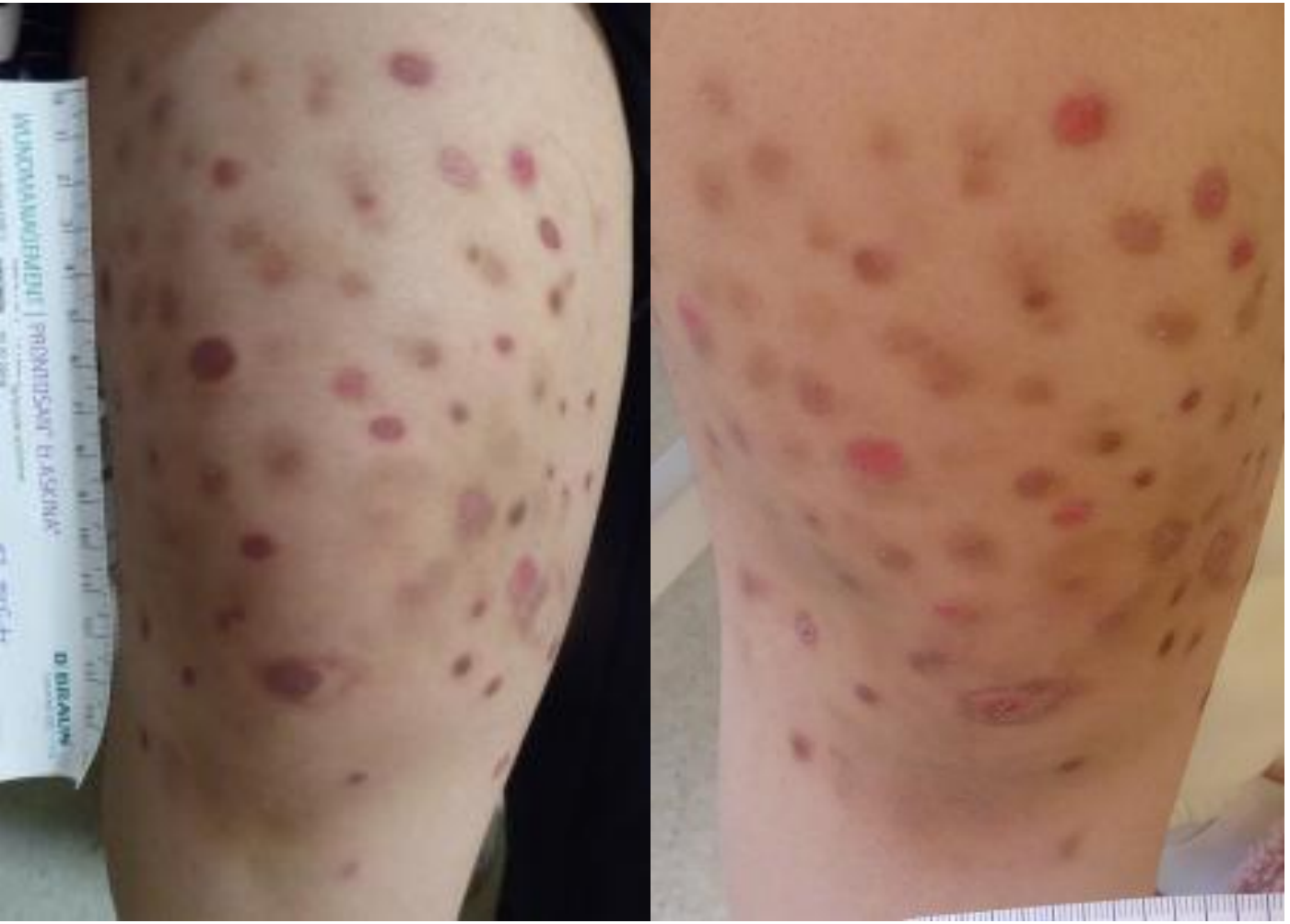


Untypical presentation…

Admission!

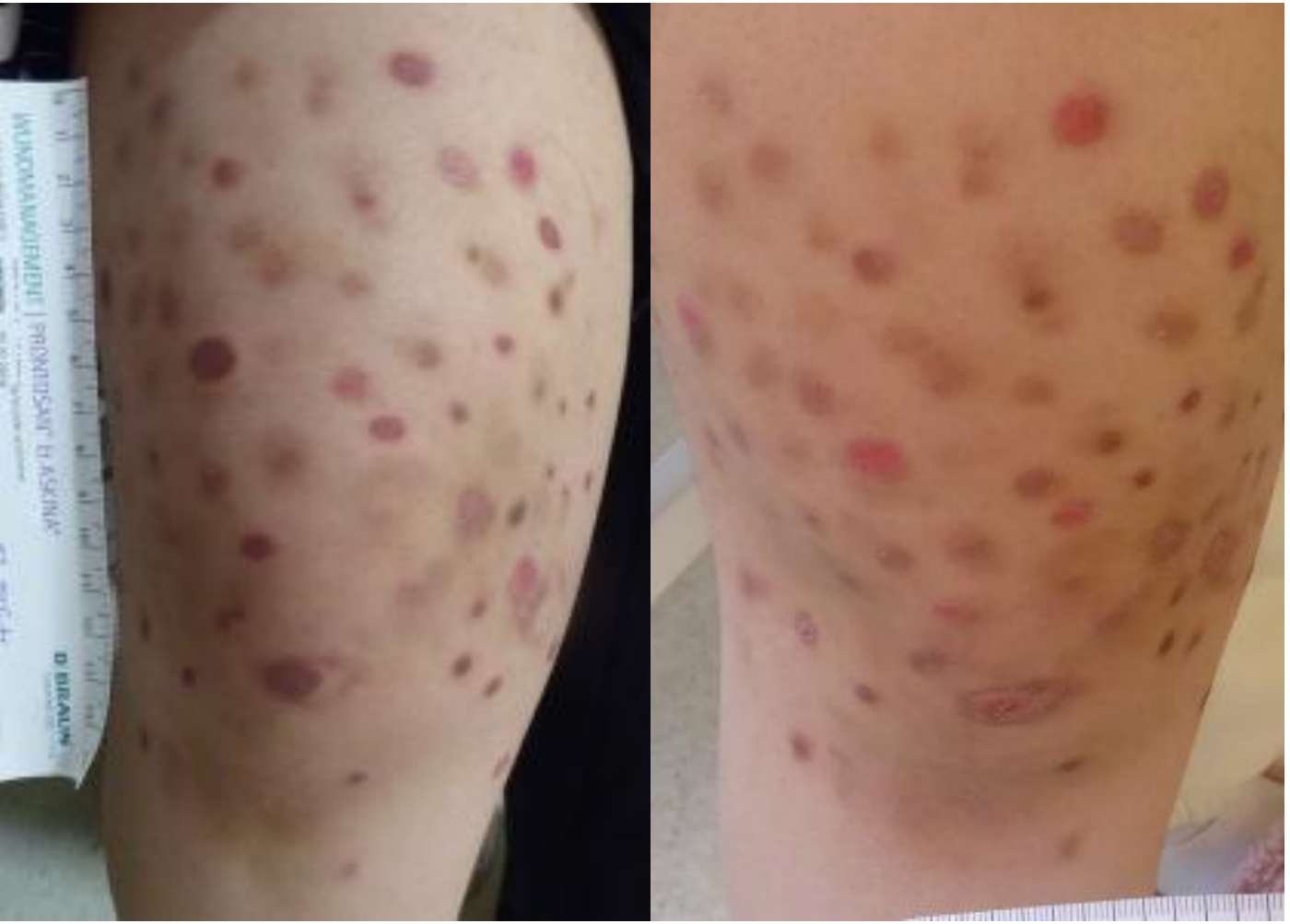
Dermatologic and pediatric work-up

- Swabs for bacteria and herpes simplex: negative
- External lesional biopsy: unspecific
- Extensive blood work, coagulation diagnostic: normal

Psychiatric evaluation

- Depressive episode, anxiety
- Continuous psychological counselling rejected

Dermatitis artefacta



Lesions

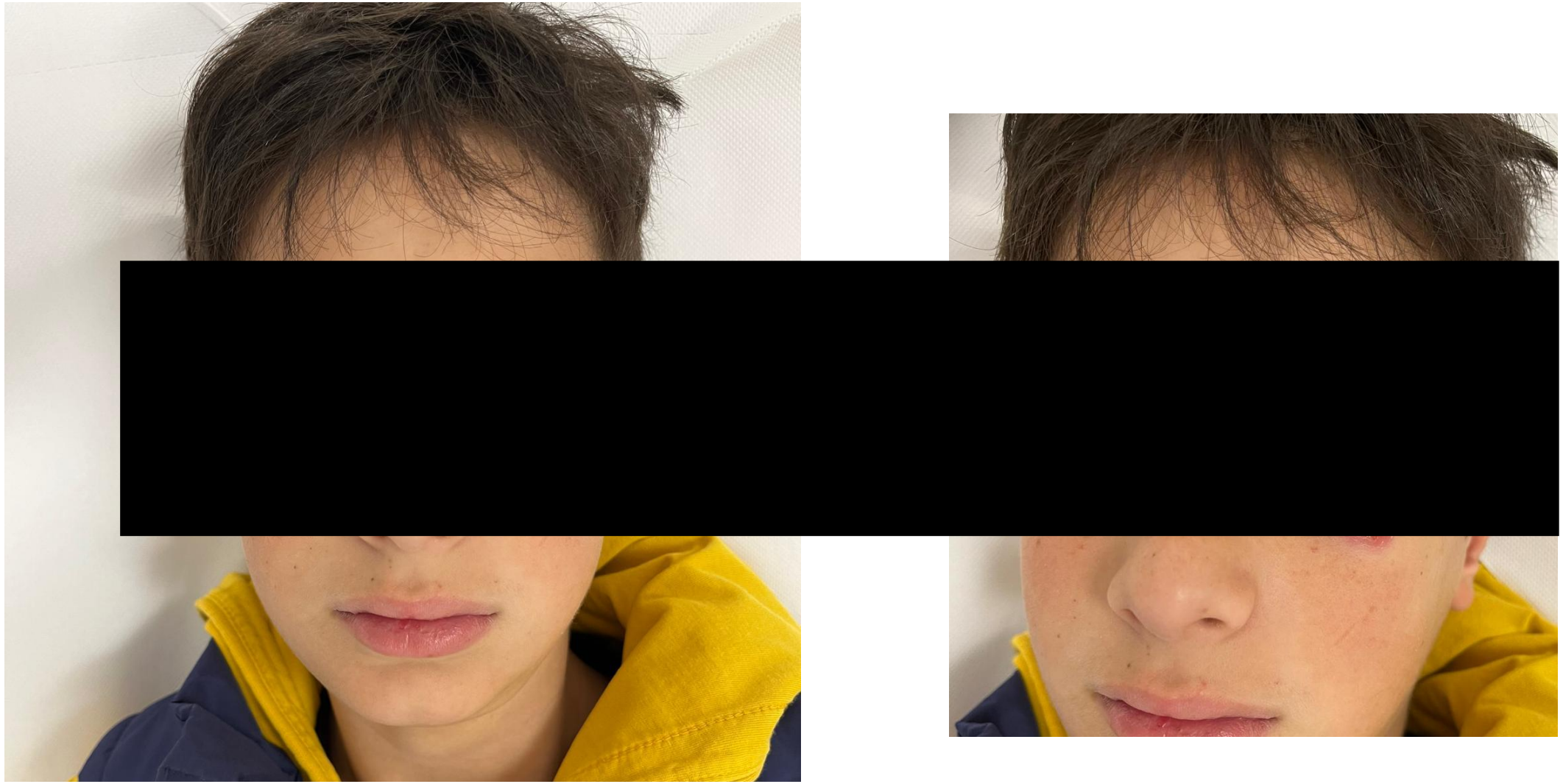
- bizarrely configured, geometric, unilateral
- within reach of the hands
- sudden appearance of complete lesions

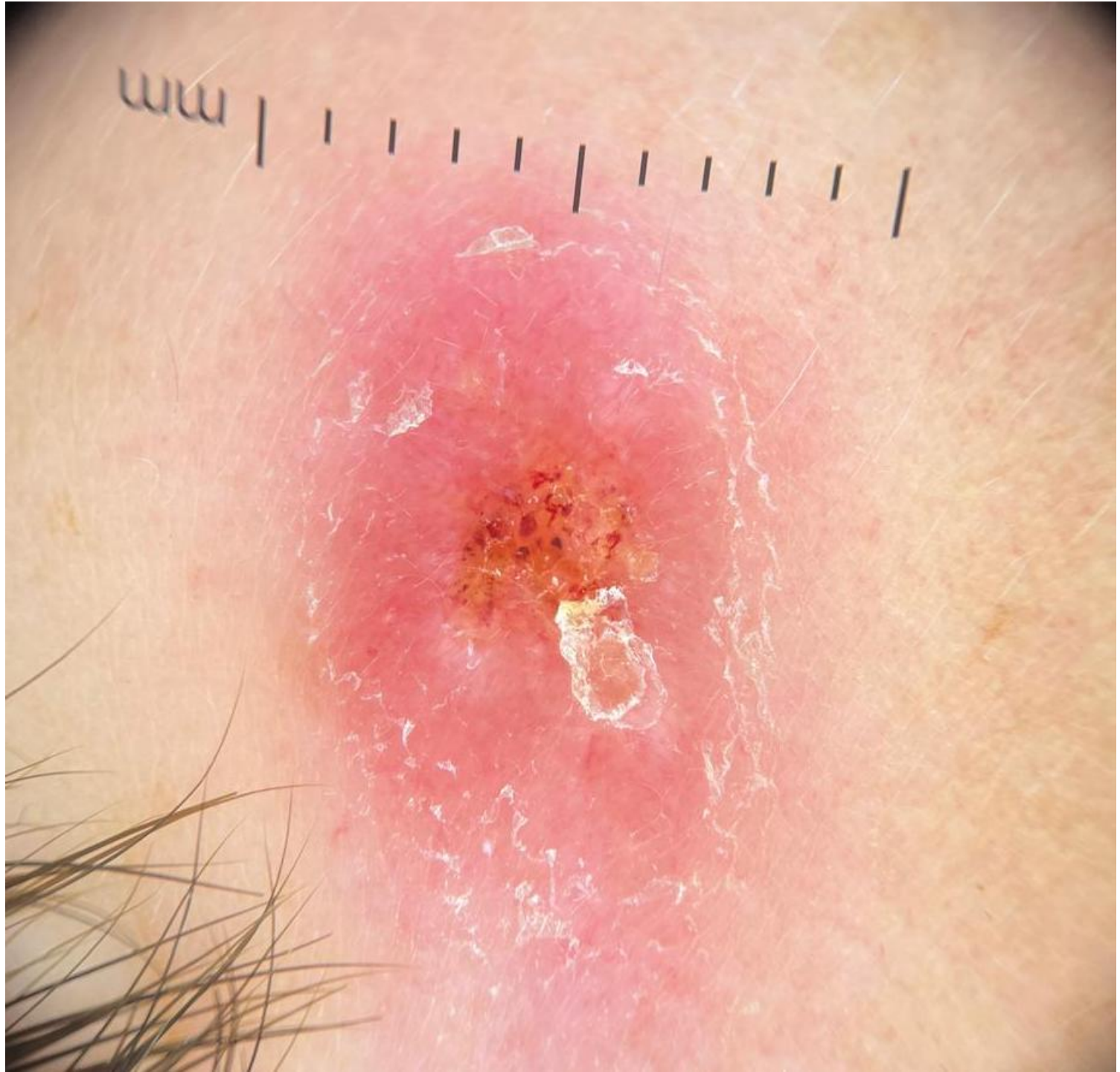
Mechanisms

- irritating or toxic substances
- mechanical injuries (crushing, suction) to cause purpura, hematomas, excoriations, ulcers
- In toddlers: by proxy

Therapy: challenging

13-year-old patient with a lesion present for approximately 2 months.
Unresponsive to topical and systemic antibiotic therapy.
The patient reports no symptoms.





Diagnosis

1 – LEISHMANIASIS

2 – DESMOPLASTIC SPITZ NEVUS

3 – ULCERATED PILOMATRIXOMA

4 – TRICOBLASTOMA

5 – LYMPHOCYTOMA

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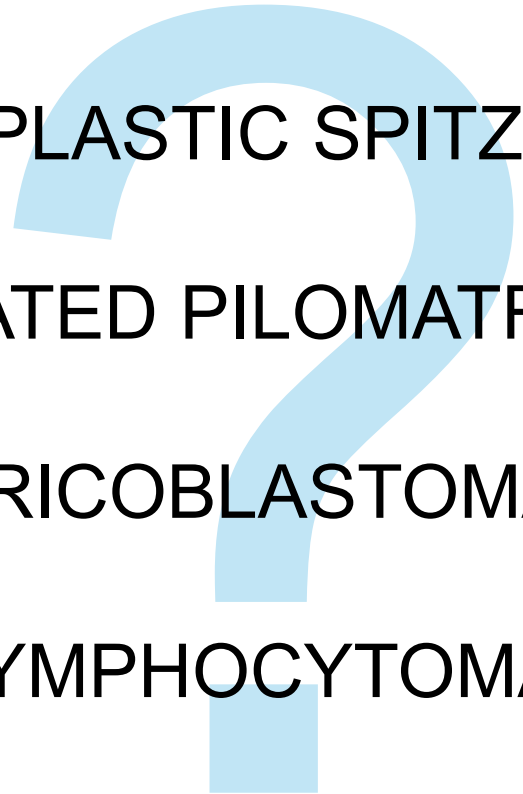
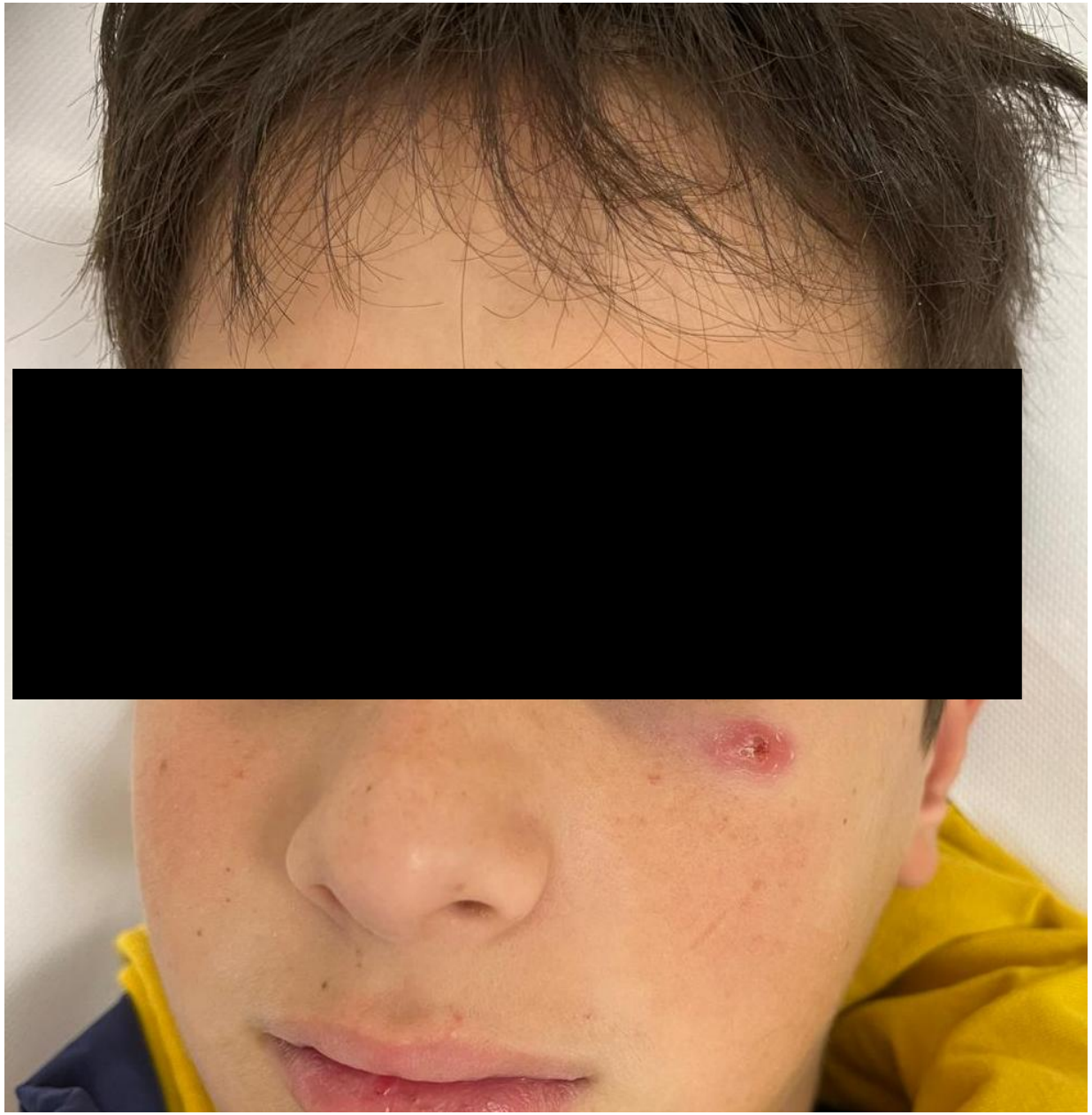
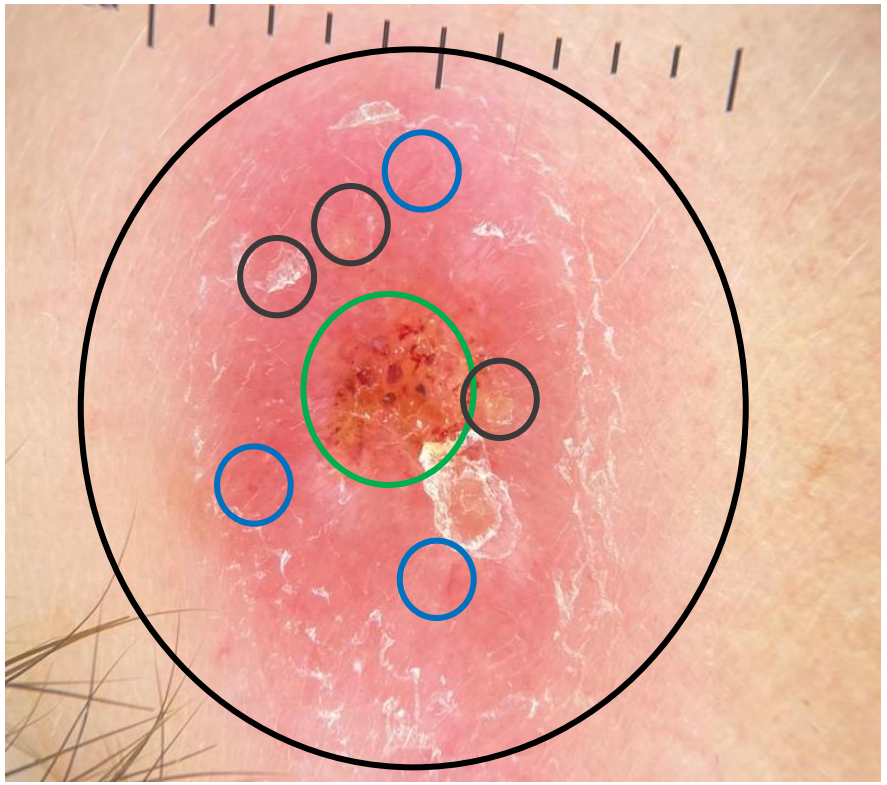


CUTANEOUS LEISHMANIASIS





- Erithema
- Comma-shaped vessels
- Yellow-tears
- Ulceration and hyperkeratosis

Dermatology Practical & Conceptual

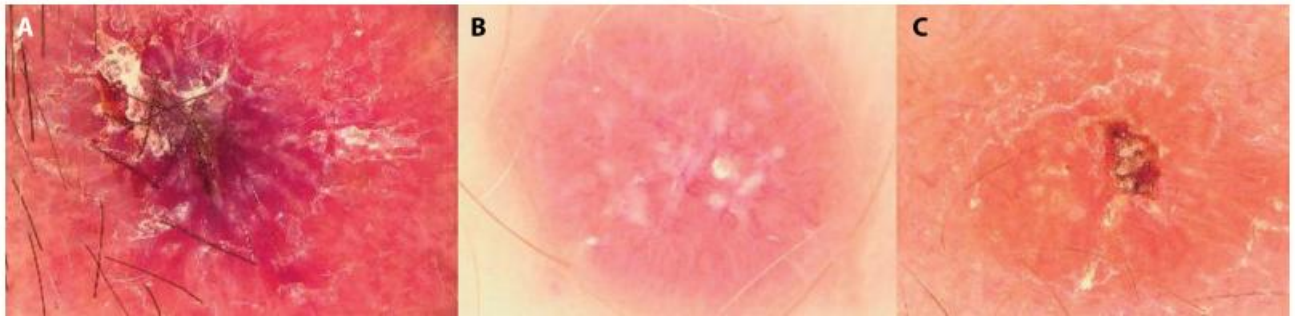
Dermoscopy in the Diagnosis of Cutaneous Leishmaniasis

Prospective Study on 79 Patients (Turkey)
Erythema in **100%** of cases
Yellow tears in **75.5%**
White starburst pattern in **58.3%**
Vascular structures in **94.2%**, with the most common being:
- Irregular linear vessels (45.8%)
- Hairpin vessels (43.5%)
- Comma-shaped vessels (25.9%)
- Arborizing vessels (18.3%)

Correlations
Limbs → hyperkeratosis and starburst pattern
Head–neck → arborizing vessels
Duration >6 months → micro-arborizing vessels

Table 2. Dermoscopic Features in CL

Dermoscopic Features	n (%)
General features	
Erythema	139 (100)
Dusky red	47 (33.8)
Light red	61 (43.9)
Yellowish red	31 (22.3)
Hyperkeratosis	49 (35.3)
CE/U	10 (7.2)
HK+E/U	23 (16.5)
Yellow tears	105 (75.5)
White starburst-like pattern	81 (58.3)
White scar-like patch	7 (5.0)
Milia-like cyst	12 (8.6)
Yellowish hue	1 (0.7)
Salmon-colored ovoid	4 (2.9)
Vascular features	131 (94.2)
Comma-shaped	34 (25.9)
Linear irregular	60 (45.8)
Dotted	19 (14.5)
Hairpin	57 (43.5)
Arborizing	24 (18.3)
Corkscrew	5 (3.8)
Glomerular-like	28 (21.4)
Milky red globules/areas	15 (11.4)
Microarborizing	21 (16.0)
Crown	4 (3.0)



Serarslan G et al. Dermoscopy in the Diagnosis of Cutaneous Leishmaniasis. Dermatol Pract Concept. 2019 Apr 30;9(2):111-118

ORIGINAL PAPER

Dermoscopic evaluation of cutaneous leishmaniasis

Muhsin A. Al-Dhalimi[1] · Shadan Hussein Jasim[2]

Table 1 Dermoscopic features of the CL lesions

Dermoscopic features	No	%
Erythema	91	100
Dusky red	29	31.9
Light red	53	58.2
Yellowish red	9	9.9
HK + E/U	49	53.8
White scar-like patch	38	41.8
Yellow tears	32	35.2
Hyperkeratosis	32	35.2
White starburst-like pattern	31	34.1
Yellowish hue	11	12.1
CE/U	9	9.9
Hypopigmented halo	3	3.3
Milia-like cyst	2	2.2
Salmon colored ovoid	0	0
Vascular features	84	92.3
Linear irregular	53	63.1
Dotted	48	57.1
Glomerular	32	38.1
Hairpin	19	22.6
Comma shaped	14	16.7
Milky red areas	13	15.5
Micro arborizing	10	11.9
Arborizing	6	7.1
Corkscrew	5	6
Crown	0	0

Osservational Cross-Sectional Study (Iraq, 2019–2020) – 67 Patients, 91 Lesions

- **Erythema** observed in **100%** of lesions, frequently associated with:
  - **Hyperkeratosis / erosion or ulceration** (53.8%)
  - **White scar-like patches** (41.8%)
  - **Yellow tears** (35.2%)
  - **White starburst-like pattern** (34.1%)
- **Vascular structures** identified in **92.3%** of cases:
  - Irregular linear vessels (63.1%)
  - Dotted vessels (57.1%)
  - Glomerular vessels (38.1%)
  - Hairpin vessels (22.6%)
  - Comma-shaped vessels (16.7%)
  - Arborizing vessels (7.1%)

Correlations

- **Face / upper limbs** → irregular linear vessels
- **Lower limbs** → hyperkeratosis + ulceration
- **Papular pattern** → hyperkeratosis / ulceration
- **Nodular pattern** → irregular linear vessels
- **Nodulo-ulcerative pattern** → glomerular vessels (**p=0.02**)
- **Plaque lesions** → irregular linear vessels (71%)

Al-Dhalimi MA et Dermoscopic evaluation of cutaneous leishmaniasis. Arch Dermatol Res. 2023 Apr;315(3):531-540

THERAPY

Galenic formulation containing:
•Paromomycin sulfate 15%
•Urea 6.75%
•Gentamicin 0.5%

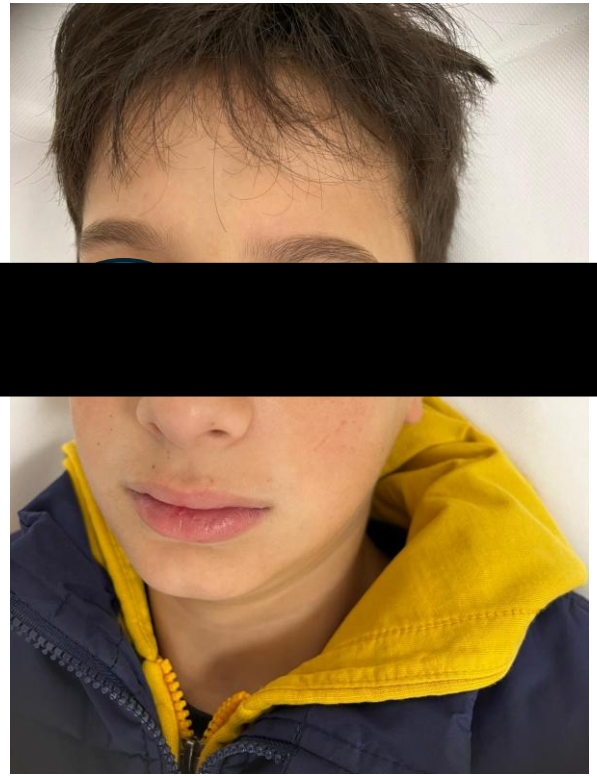
Twice daily for 2 months

Ben Salah A PLoS Negl Trop Dis. 2009;3(5):e432.

W0

W4

W8



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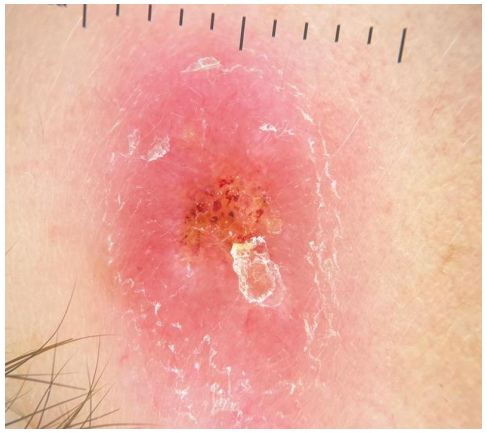

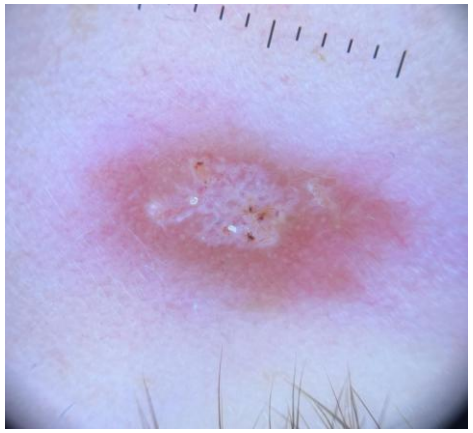

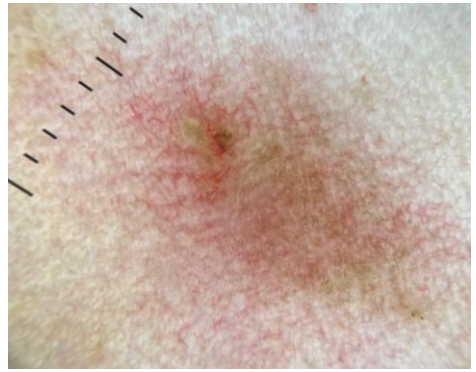

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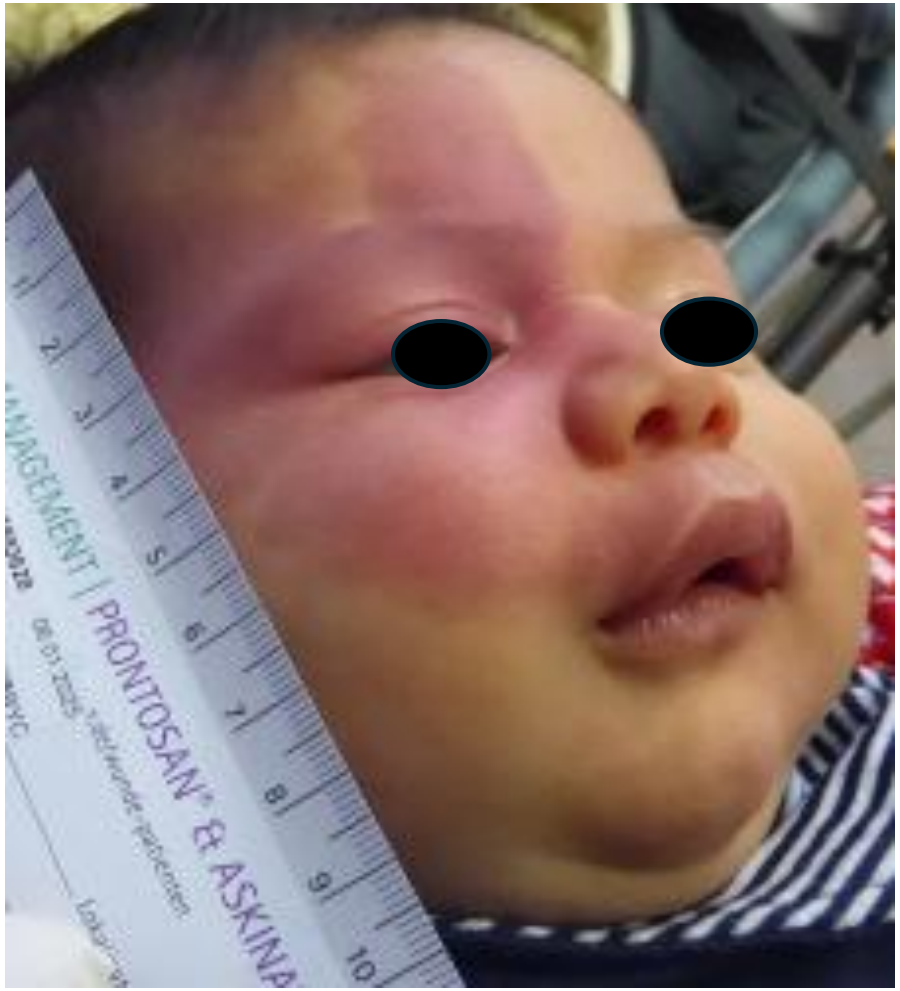
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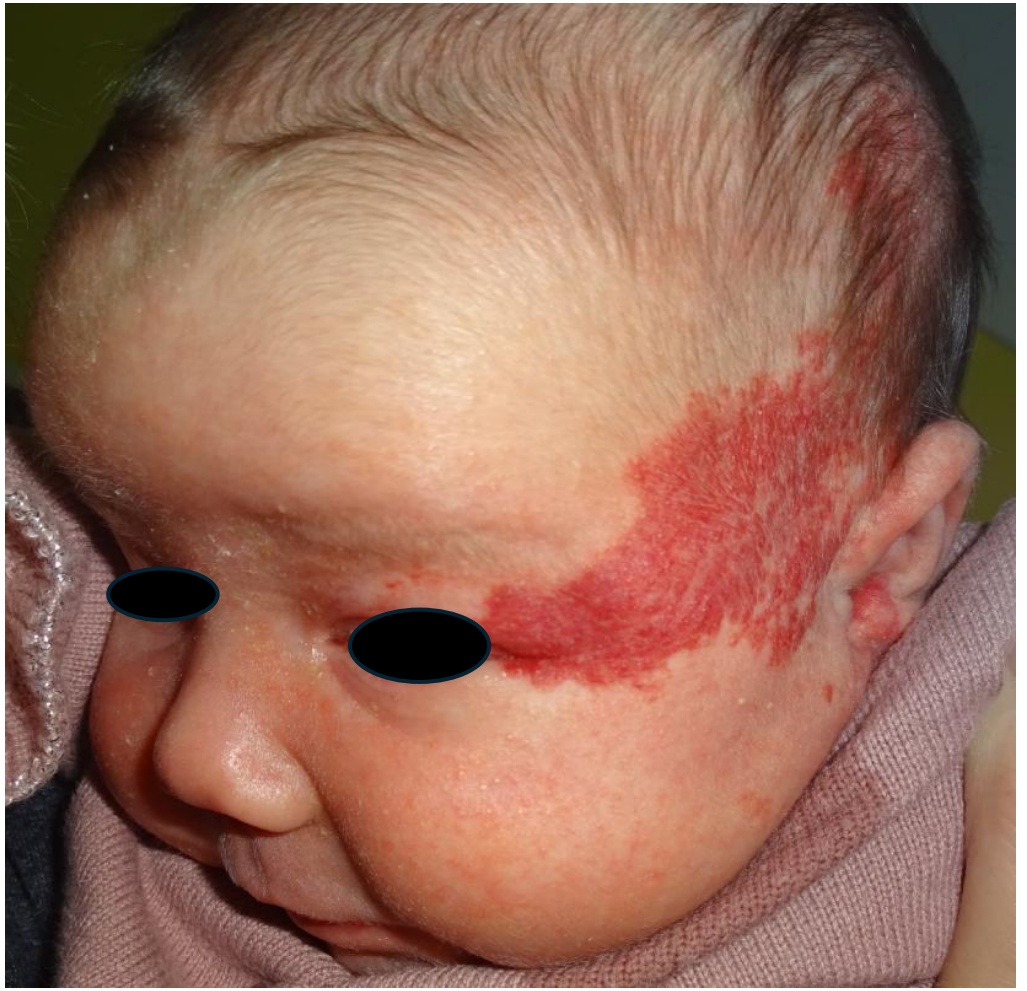
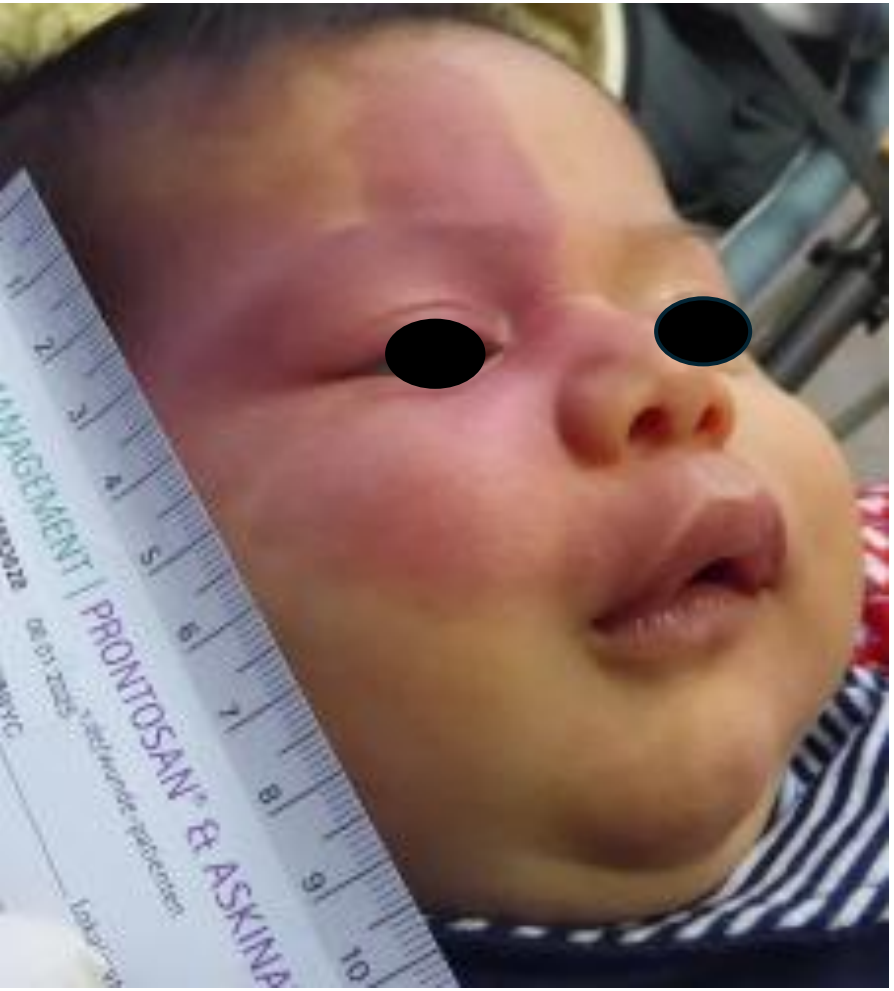


Sturge - Weber syndrome



Diagnostic work-up

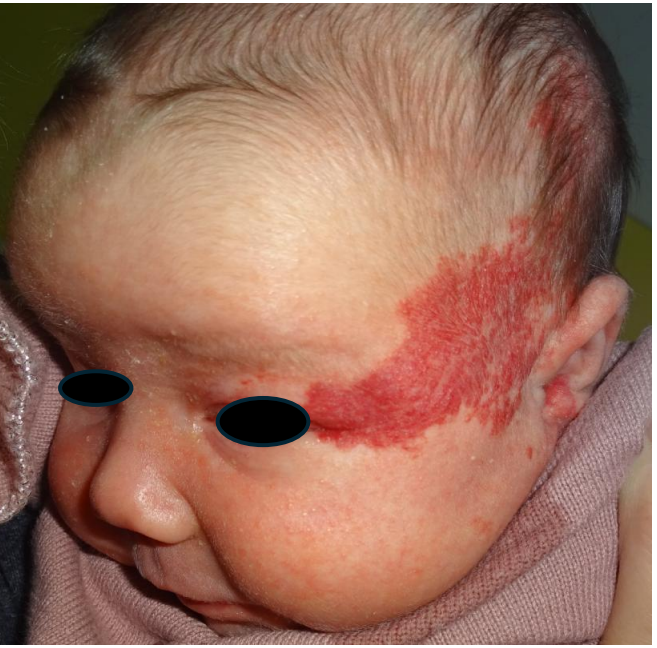
- MRI: increased leptimeningeal contrast enhancement parieto occipital, prominent right plexus choroideus
- no sign of ocular involvement

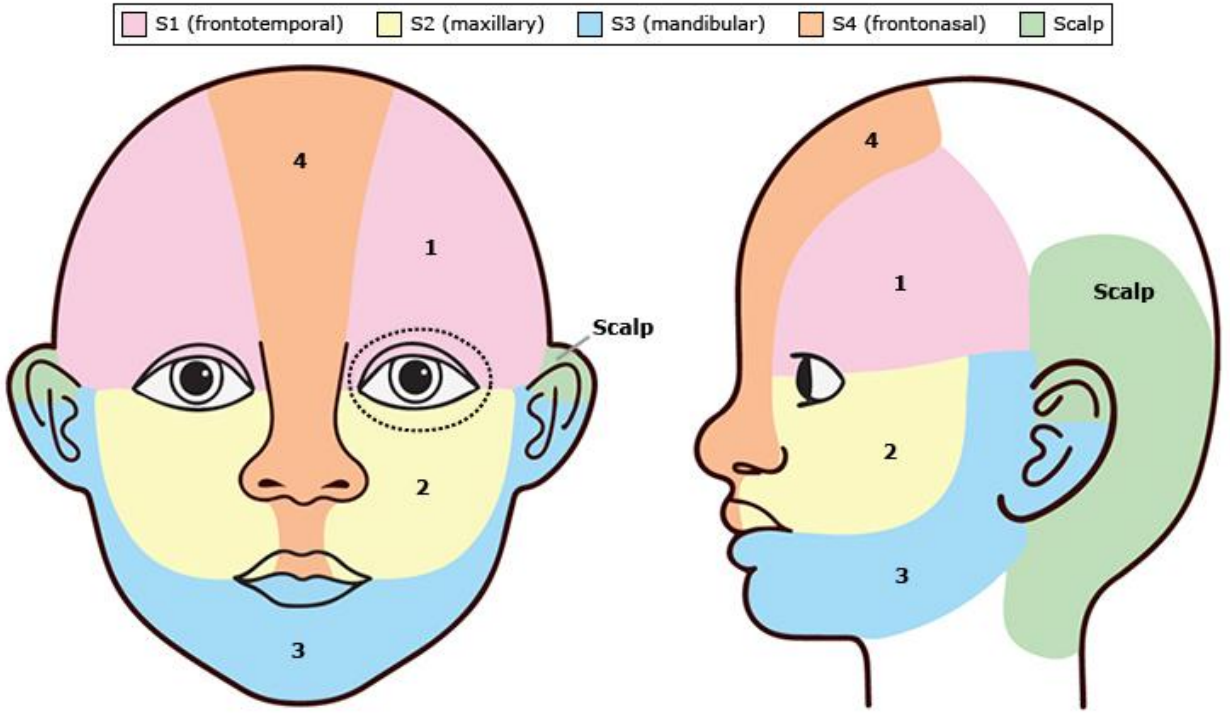
Treatment

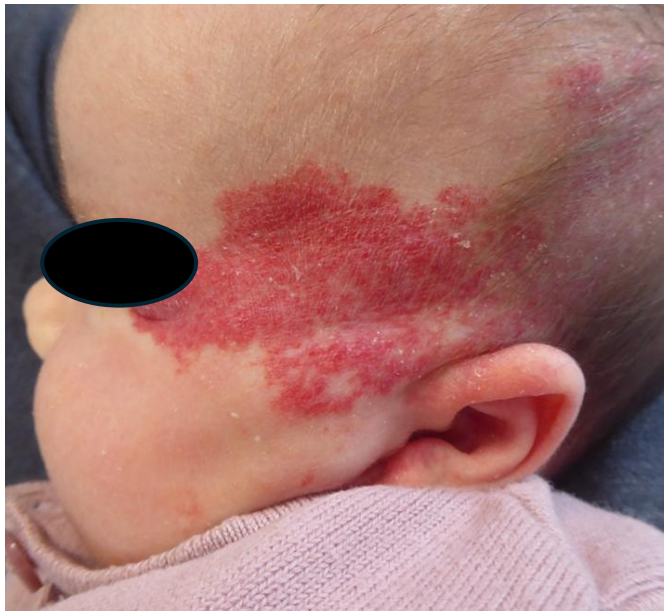
- antiepileptic (Oxcarbazepine), acetylsalicylic acid low-dose
- (future: Sirolimus)
- Laser

El Hachem *et al. Orphanet J Rare Dis*. 2025

PHACE syndrome

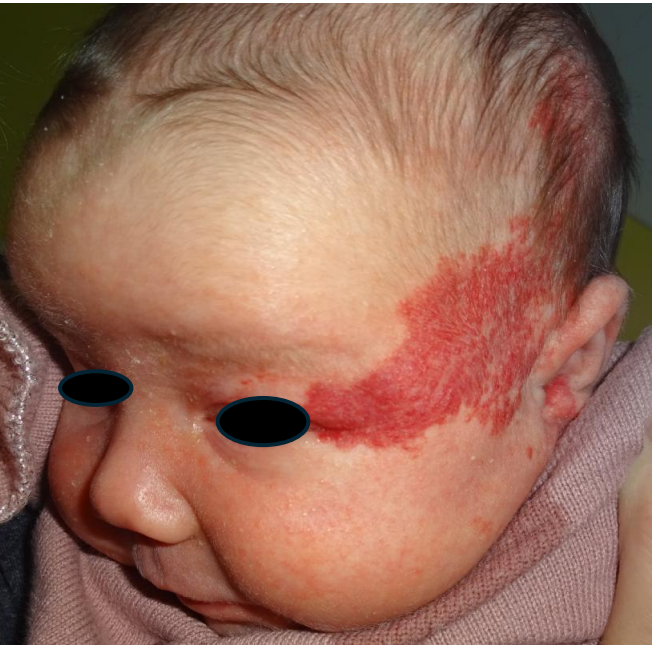


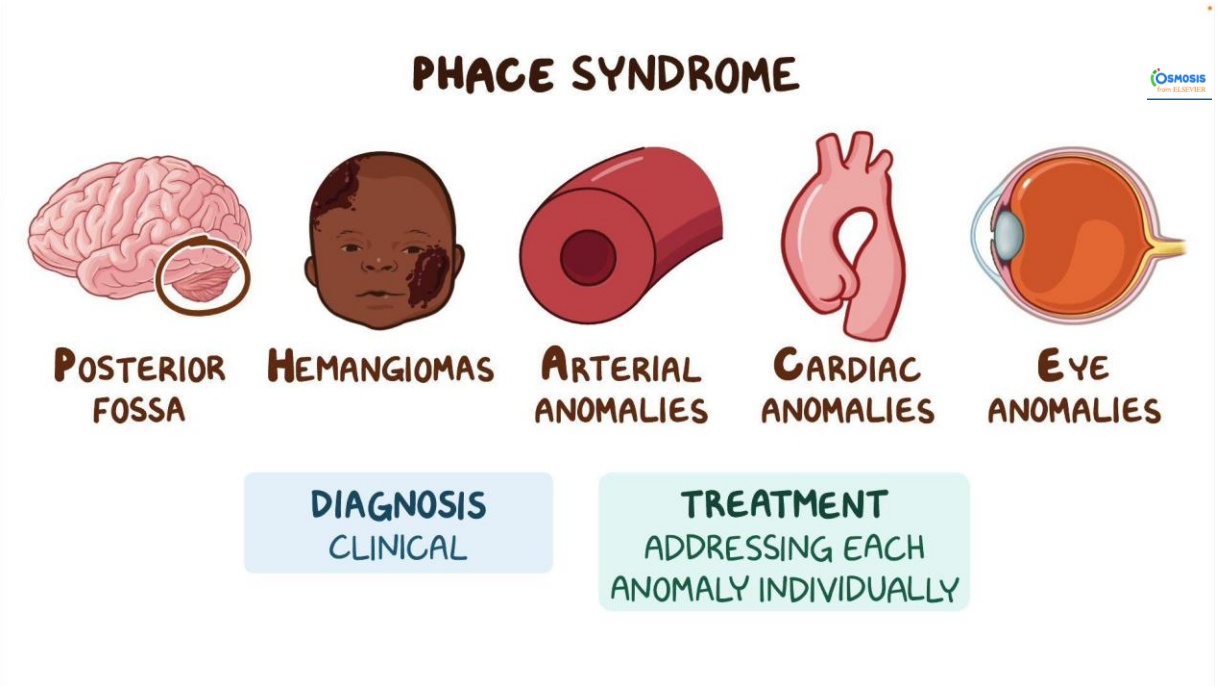


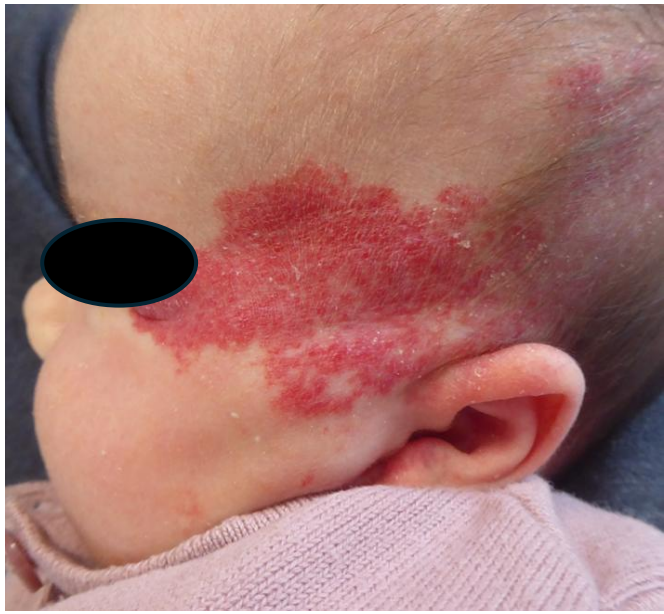


Endicott AA *et al*. *JAMA Dermatol.* 2021

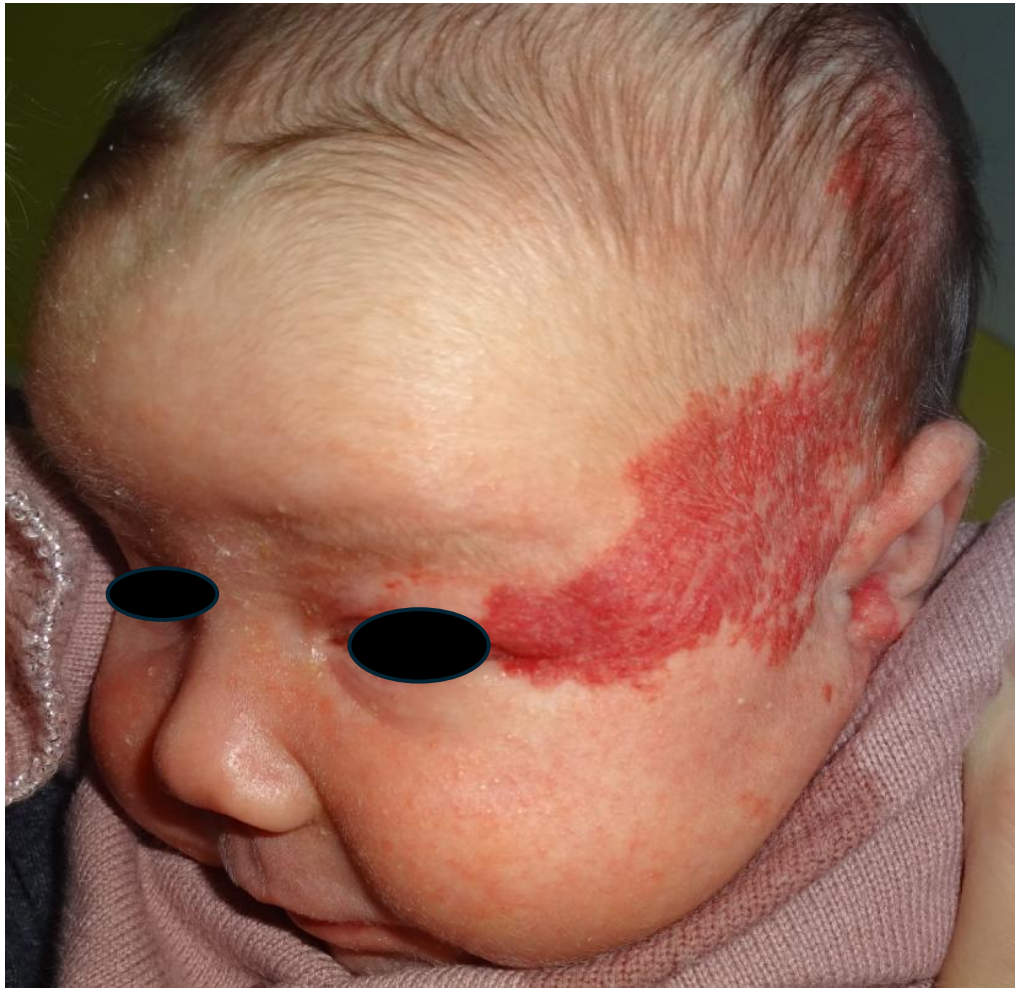
PHACE syndrome



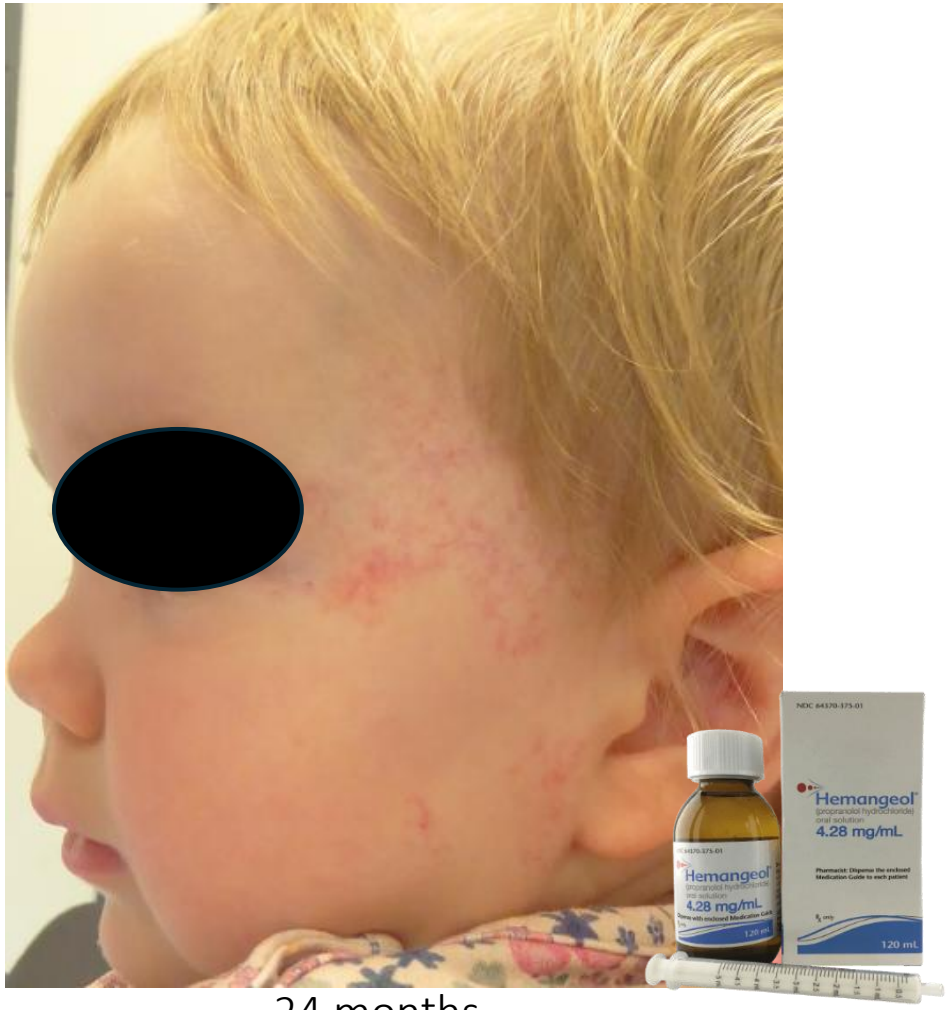




PHACE syndrome



4 weeks



24 months

Female neonate born at term via cesarean section
Non-consanguineous parents; uncomplicated pregnancy
Birth weight 2530 g; Apgar score 9/10
Good general clinical conditions at birth

Immediately after delivery, the birth center requested a consultation for suspected epidermolysis bullosa

Ambra, 3 days old

Serous-filled bullous lesions measuring approximately 2 cm, along with erosions on the right upper limb and left lower limb.

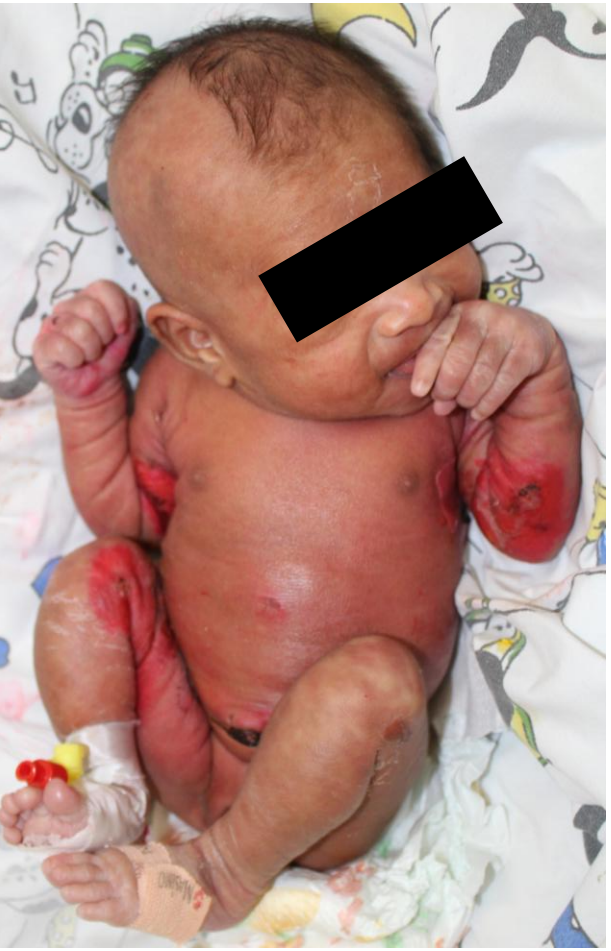
Erythroderma

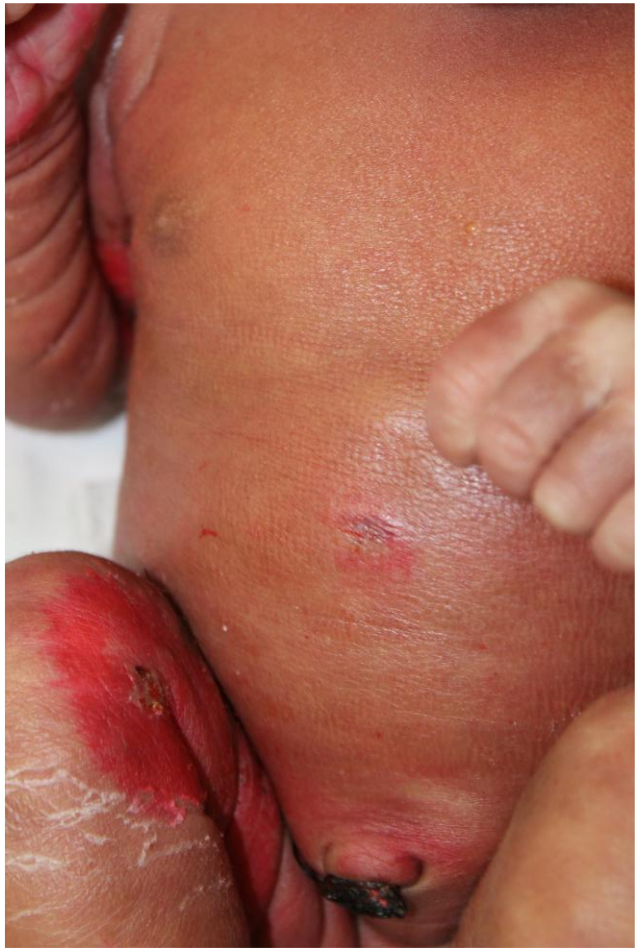
Redundant skin with accentuated skin folds, markedly pasty and thickened ("pachydermic") on both upper and lower limbs
Trunk shows tight skin with accentuated dermatoglyphics, without signs of laxity

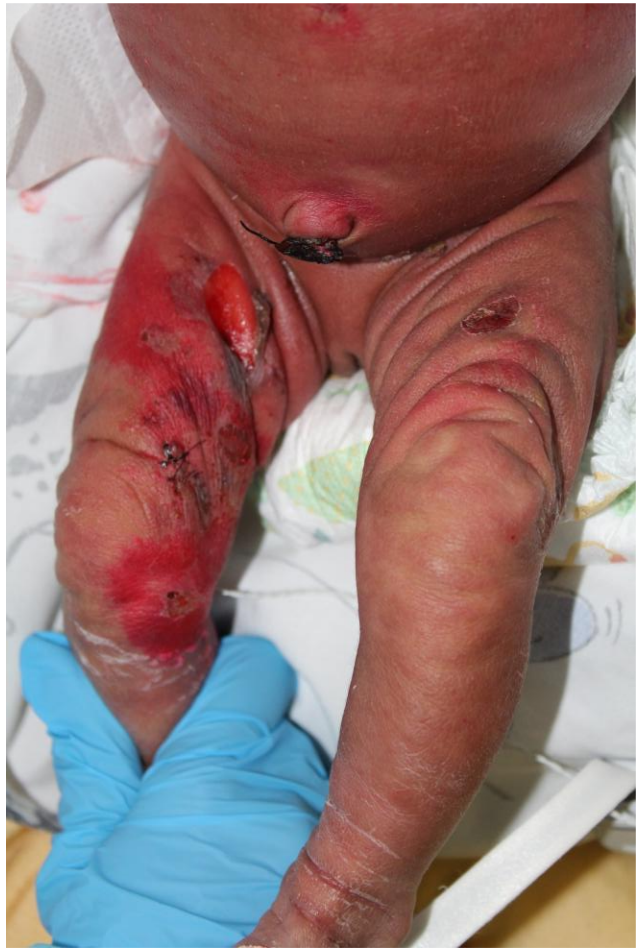
Limb erosions resulting from serous bullae, not surrounded by erythematous halo

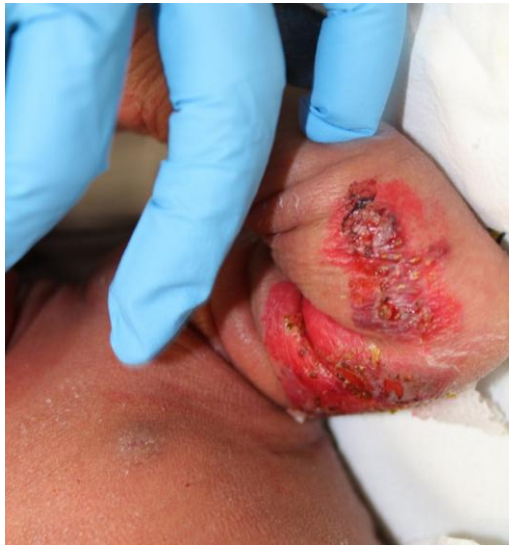
Lesional and perilesional skin biopsy for histological examination, immunofluorescence, and ultrastructural analysis.

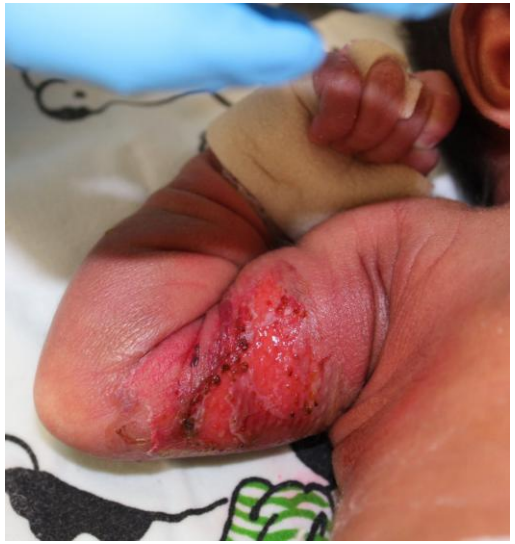
Ambra, 5 days old









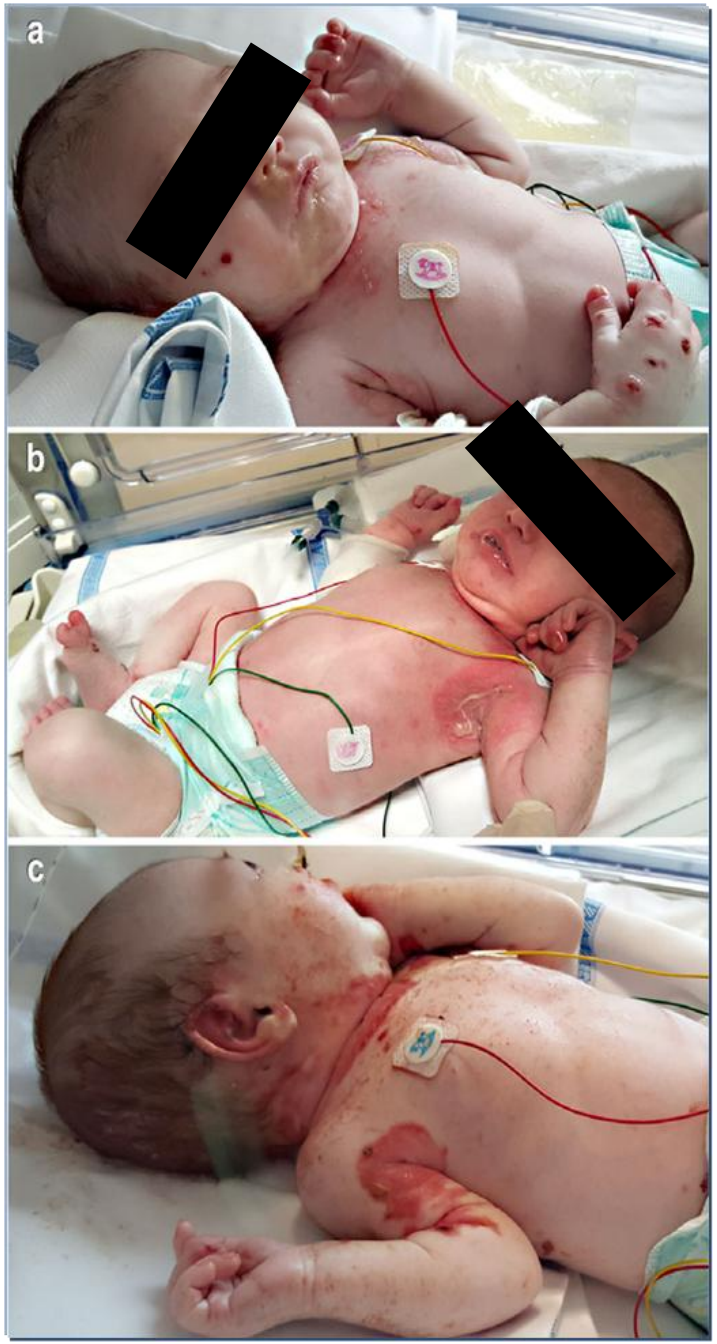


Diagnosis

1. Inherited epidermolysis bullosa?
2. Neonatal linear IgA bullous dermatosis?
3. Cutis laxa?
4. Storage disorders?
5. Other?

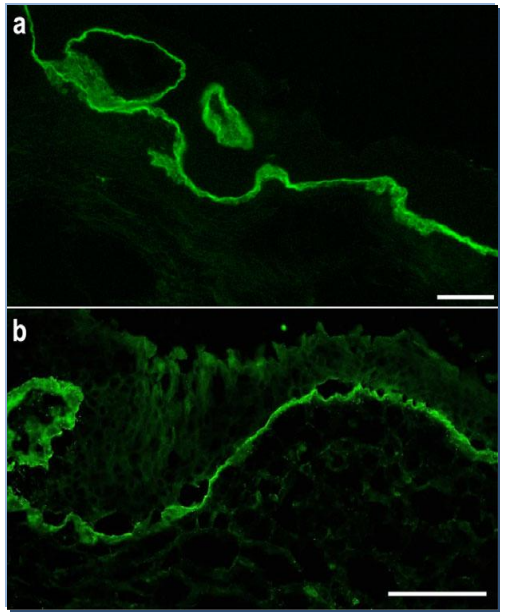
Diagnosis

1. Inherited epidermolysis bullosa?
2. Neonatal linear IgA bullous dermatosis?
3. Cutis laxa?
4. Storage disorders?
5. **Other?**



Neonatal linear IgA bullous dermatosis? ❌

No, because:
No lesions on skin folds, oral mucosa, or ocular surfaces
Absence of respiratory symptoms

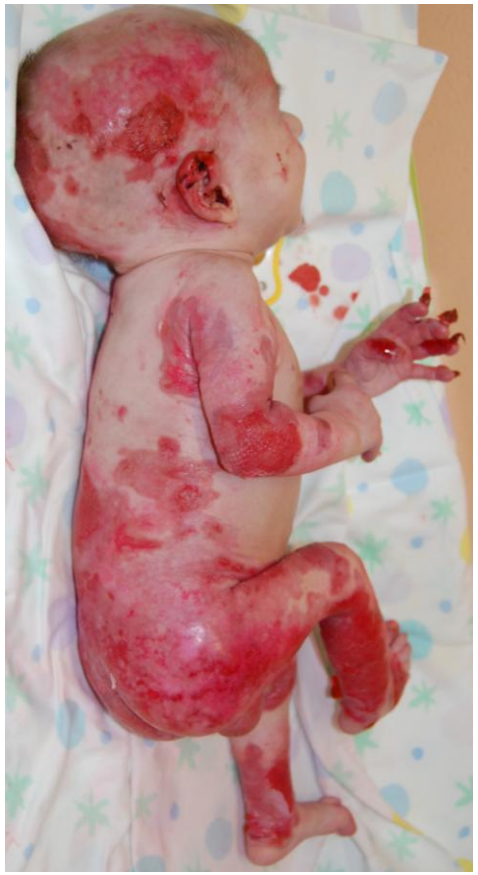


IFD:
(a) **Linear IgA deposition along the skin**
(b) **Same pattern observed in the bronchi**

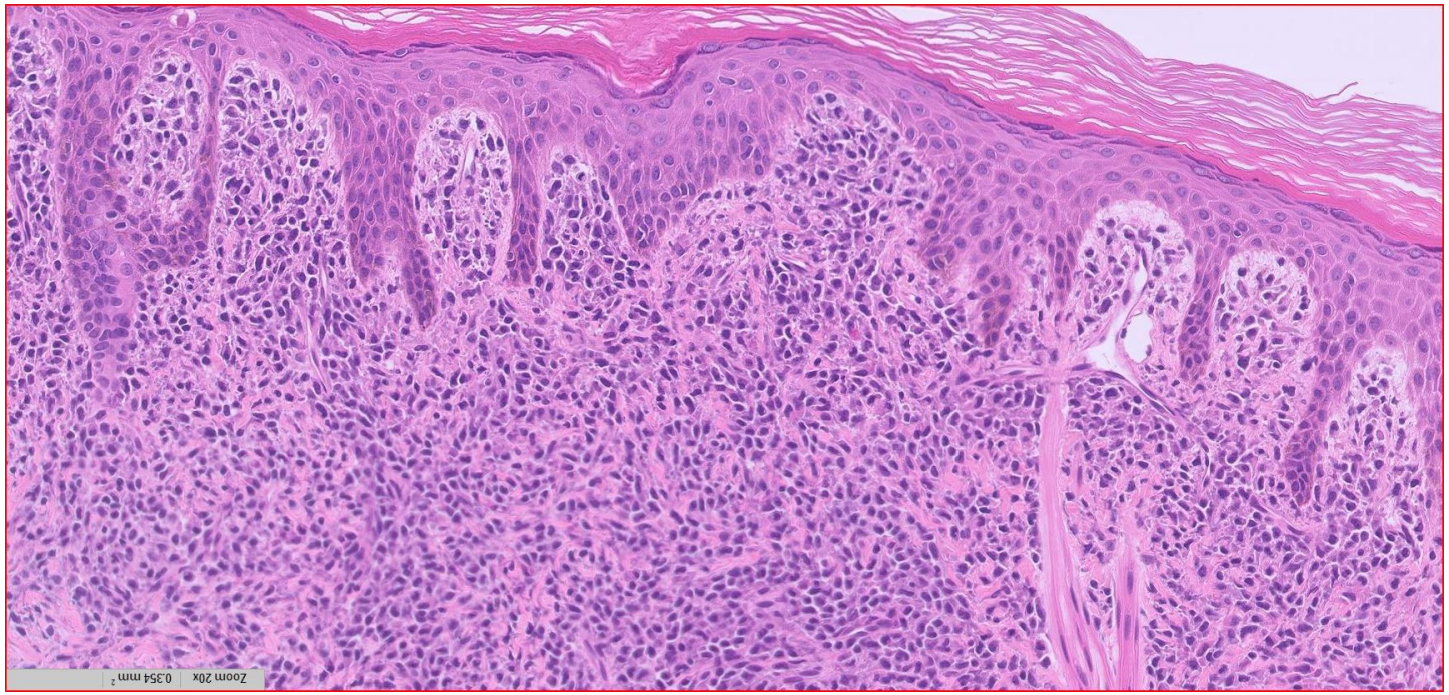
Hereditary epidermolysis bullosa?

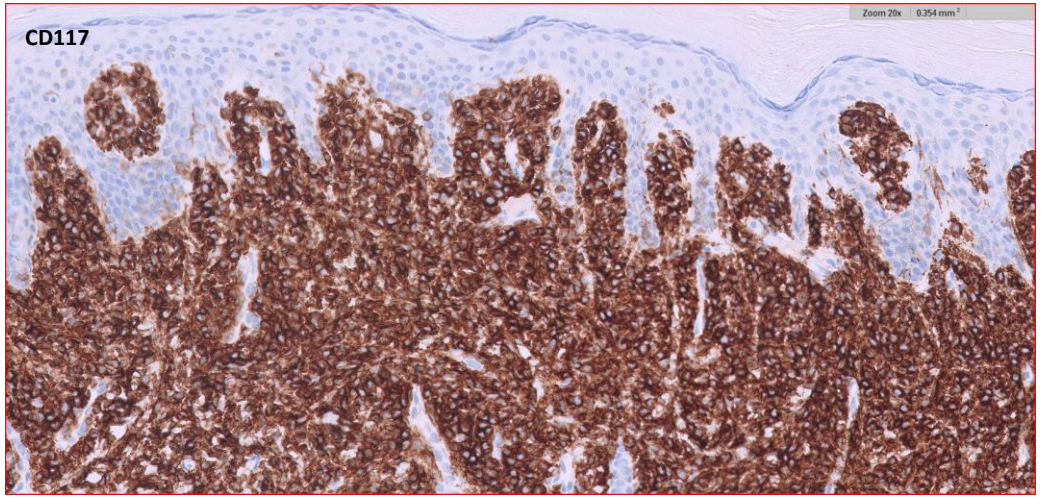
No, because:

- Vesiculobullous lesions on the skin and sometimes also in the oral cavity, with serous or hemorrhagic content
- Erosive areas of variable extent
- Absent nails or onychodystrophy
- Skin unaffected by blisters appears normal although fragile



Histologic Exam





Skin showing a dense infiltrate of monomorphic cells with amphophilic cytoplasmic granules, involving the full thickness of the dermis and focally the subcutaneous tissue.
The infiltrate tested positive for CD117 and tryptase.

DIAGNOSIS:
DIFFUSE CUTANEOUS MASTOCYTOSIS

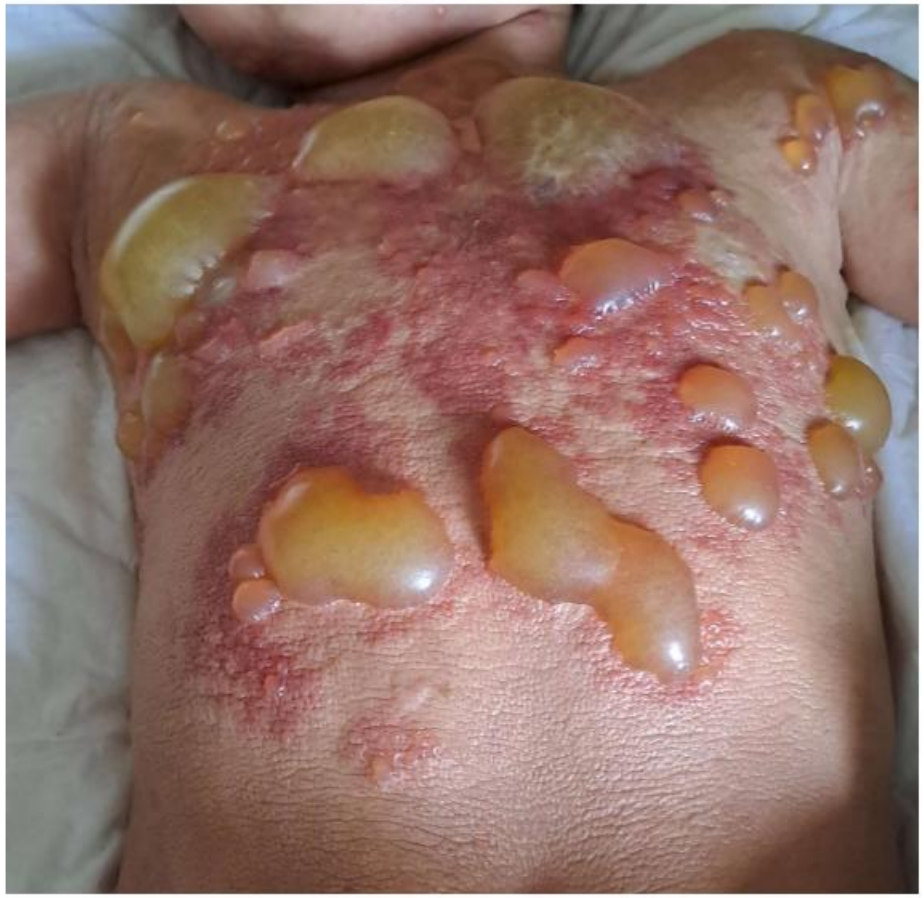
Table 1. Updated Classification of Mastocytosis.

Cutaneous mastocytosis (CM)
• Maculopapular cutaneous mastocytosis (MPCM)
○ Monomorphic variant
○ Polymorphic variant
• Diffuse cutaneous mastocytosis (DCM)
• Cutaneous Mastocytoma
Systemic mastocytosis (SM)
Non-advanced forms of SM
• Indolent systemic mastocytosis (ISM)
• Bone marrow mastocytosis (BMM)
• Smoldering systemic mastocytosis (SSM)
Advanced forms of SM
• SM with an associated hematologic neoplasm (SM-AHN)
• Aggressive SM (ASM)
• Mast cell leukemia (MCL)
Mast cell sarcoma (MCS)

Rydz A et. Diffuse Cutaneous Mastocytosis: A Current Understanding of a Rare Disease. Int J Mol Sci. 2024;25:1401.

Table 3. Skin diseases mimicking DCM.

Disease	Skin Lesions Resembling DCM	Main Clinical Features of the Disease
Staphylococcal scalded skin syndrome (SSSS)	Blistering Redness of the entire skin Desquamation of the skin	Denudation of the skin caused by exotoxin produced by phage group II strains of *Staphylococcus* species Usually presents 48 h after birth (rare in children older than six years) Culture from the site of the suspected primary infection is warranted
Epidermolysis bullosa (EB)	Generalized bullous eruptions	Genetic collagen disorder is characterized by skin fragility leading to blistering, wounds, and scarring Identification of typical gene mutations
Impetigo bullosa (IB)	Small vesicles that can grow into tense bullae and erosions	Superficial, highly contagious bacterial (*Staphylococcus aureus* and *Streptococcus pyogenes*) skin infection Pustules, blisters, and honey-colored crusted erosions Bacterial cultures can be used for confirmation of a diagnosis
Erythema multiforme (EM)	Blisters based on erythematous skin lesions	Target-like lesions present symmetrically on the extremities (especially on extensor surfaces) and spread centripetally Precipitating factors: infections, especially the herpes simplex virus, and medications Histology: vacuolar interface dermatitis with marked infiltration with lymphocytes along the dermo-epidermal junction
Atopic dermatitis	Pruritic rash, erythroderma in severe cases	A defect in the skin barrier causes xerosis. Severe pruritus In infants, edematous papules and plaques that may have vesicles or crust on the scalp, face, and extensor extremities
Langerhans cell histiocytosis	Extensive rush and blistering in infants	Clonal disease of the monocyte-macrophage system A wide spectrum of skin lesions Histology with immunophenotyping: accumulation of CD1a-positive and/or CD207-positive dendritic cells
Linear IgA bullous dermatosis	Plaques and papules with blistering	Widespread annular blisters that exhibit a predilection for the lower abdomen, thighs, and groin Direct immunofluorescence: linear IgA deposits on the basement membrane zone
Incontinentia pigment	Blistering rash	Blistering, present in the early stages of infancy, heals spontaneously Blistering stage, followed by the development of verrucous lesions verrucous lesions and hyperpigmentation Coexisting signs: hair loss (alopecia) and dental abnormalities



Rydz A et. Diffuse Cutaneous Mastocytosis: A Current Understanding of a Rare Disease. Int J Mol Sci.

DIFFUSE CUTANEOUS MASTOCYTOSIS

Diffuse cutaneous mastocytosis (DCM) is the rarest form of cutaneous mastocytosis.

The neonatal form (NDCM) is extremely rare and is characterized by epidermal infiltration by a clonal proliferation of mast cells during the perinatal period, initially without systemic involvement but with a high risk of anaphylaxis.

Pediatric patients may have mutation in the KIT gene

Erythroderma, skin thickening, blistering lesions, dermographism, and positive Darier's sign

Wang, H.J.; Lin, Z.M.; Zhang, J.; Yin, J.H.; Yang, Y. A New Germline Mutation in KIT Associated with Diffuse Cutaneous Mastocytosis in a Chinese Family. Clin. Exp. Dermatol. 2014, 39, 146–149

Rydz A, Lange M, Ługowska-Umer H, Sikorska M, Nowicki RJ, Morales-Cabeza C, Alvarez-Twose I. Diffuse Cutaneous Mastocytosis: A Current Understanding of a Rare Disease. Int J Mol

Treatment

Inform the family on the factors that may exacerbate the condition, causing blisters or anaphylaxis: friction, heat sources, sudden changes in temperature, fever, teething, vaccination; also, avoid irritant agents in the child's skin care.

Apply topical antibiotics and antiseptics to infected or eroded areas, respectively.

Avoid prolonged use of topical or systemic steroids.

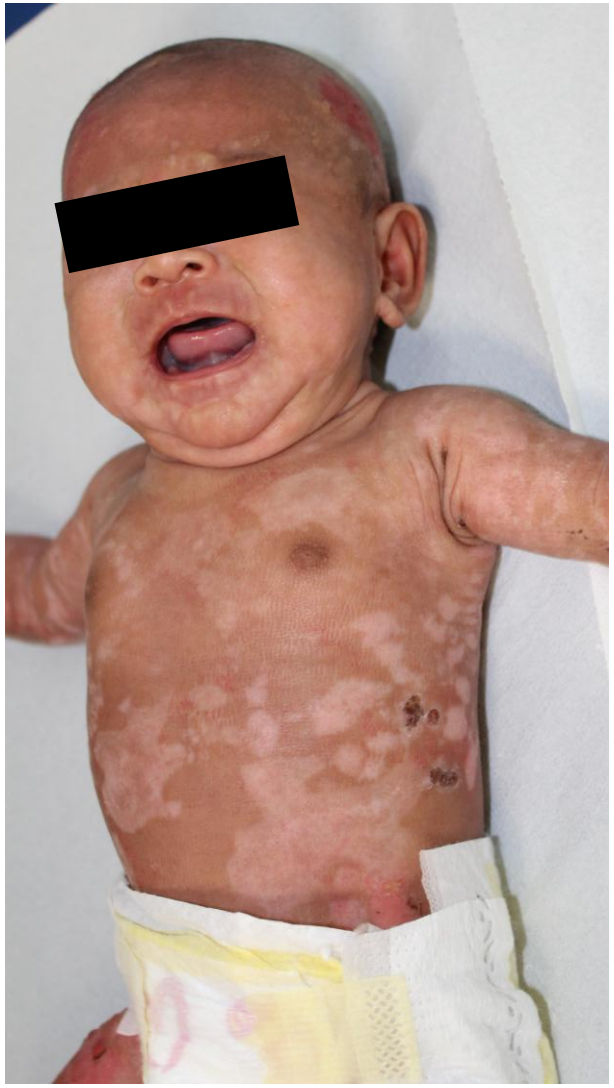
Use first-generation antihistamines, especially in children with intense itching.
Second-generation antihistamines may be used at doses up to four times higher than normal.
Sodium cromoglycate.
Phototherapy.

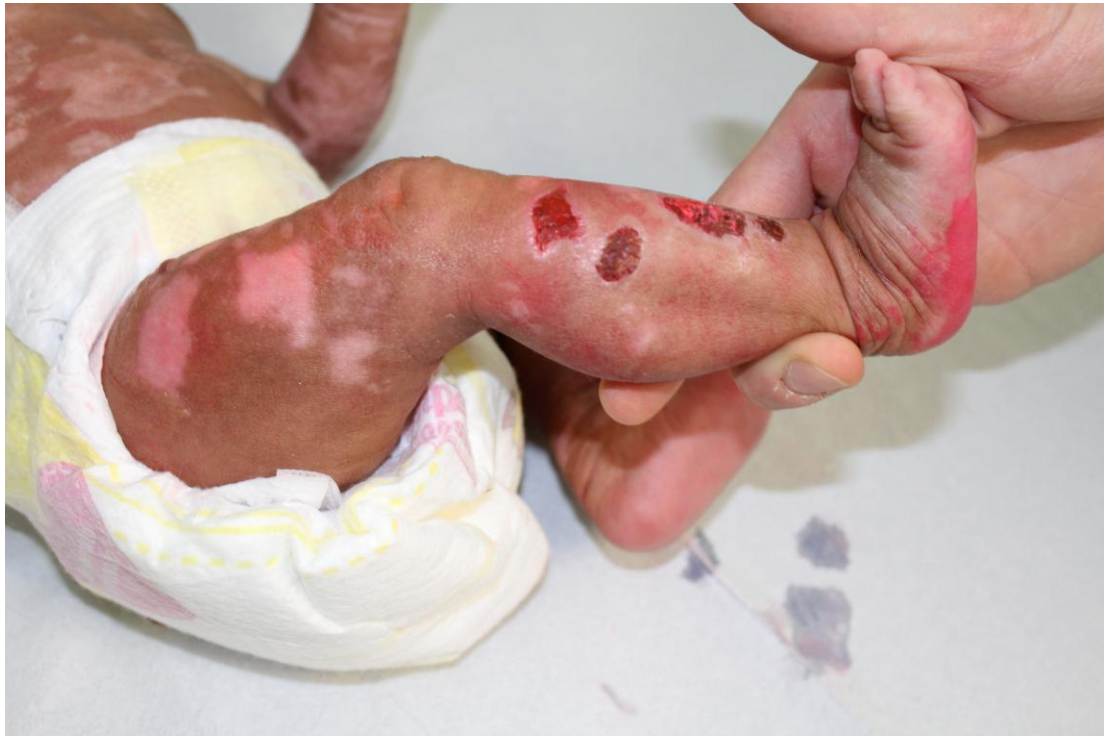
New drugs: tyrosine kinase inhibitors (imatinib in patients with KIT gene mutation), omalizumab, sirolimus.

Given the high risk of anaphylaxis, parents should be trained in the use of the adrenaline auto-injector.

Rydz A, Lange M, Ługowska-Umer H, Sikorska M, Nowicki RJ, Morales-Cabeza C, Alvarez-Twose I. Diffuse Cutaneous Mastocytosis: A Current Understanding of a Rare Disease. Int J Mol Sci. 2024;25:1401.

Ambra, 7 months





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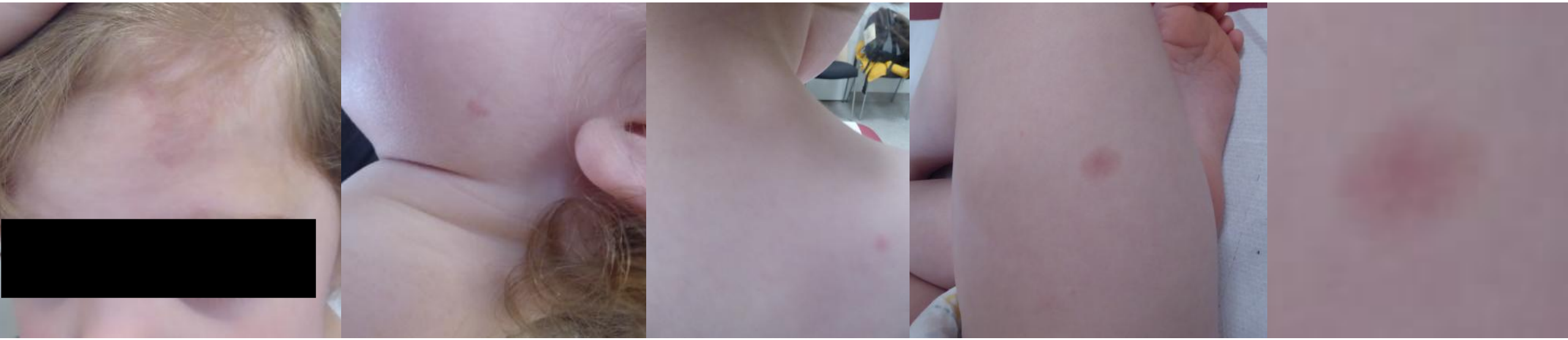

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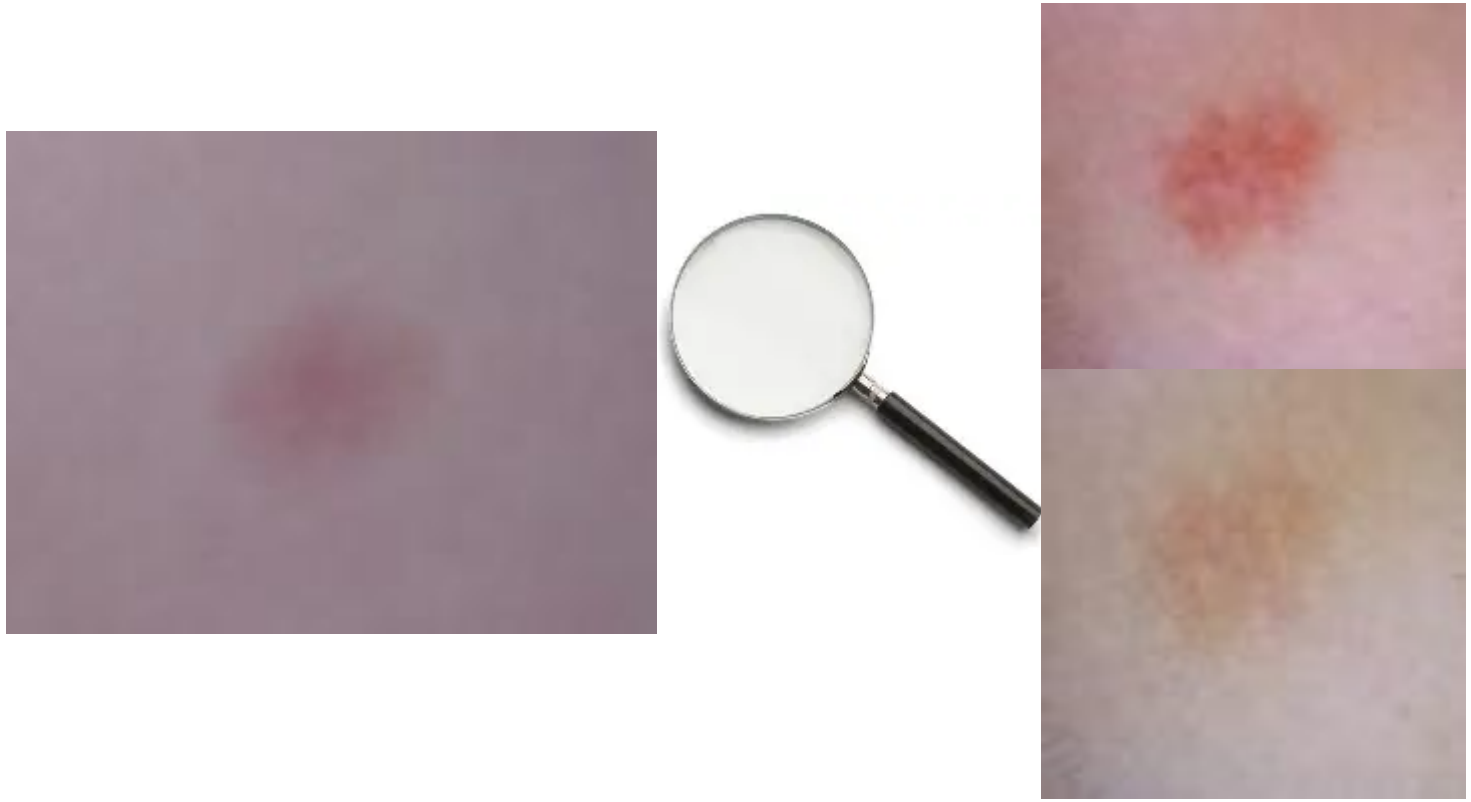

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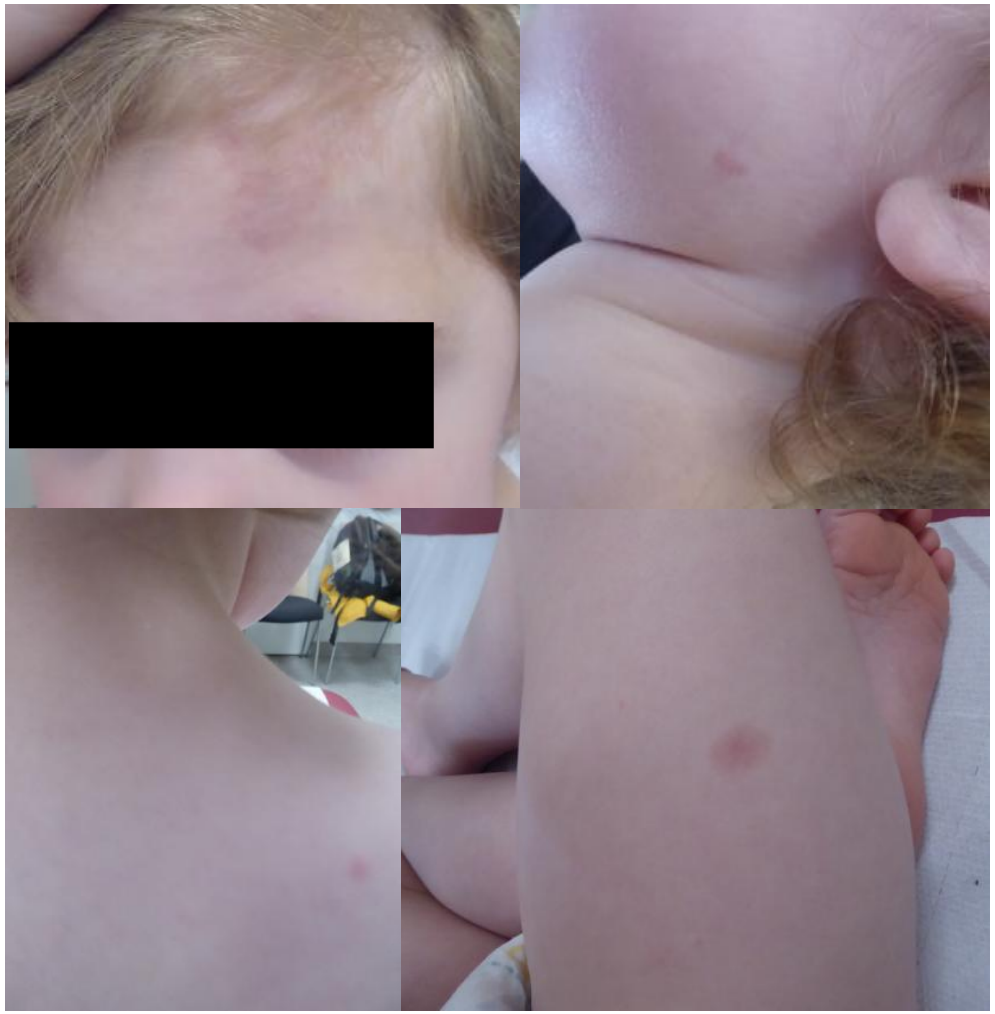


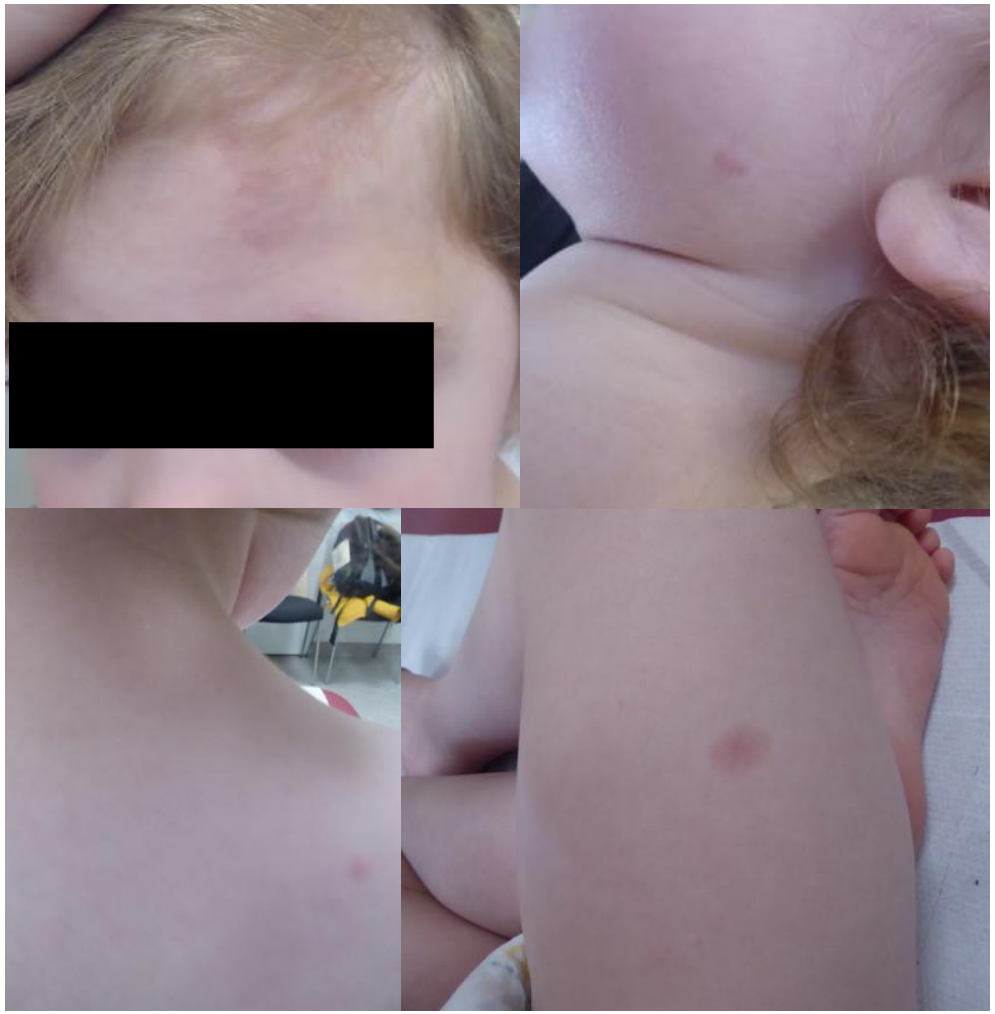
Ultrasound of skin lesions: capillary malformation, no sign of deeper malformation

Ultrasound of abdomen: no malformations

MRI brain + spine: no malformations

Genetic analysis: heterozygous *RASA1* variant

Capillary malformation – arteriovenous malformation syndrome (CMAVM)



Capillary malformations and arteriovenous malformations of skin and other tissues

Clinical presentation very variable

Causing mutations:
RASA1 (type 1) or *EPHB4* (type 2)

Hydrops fetalis can be first symptom

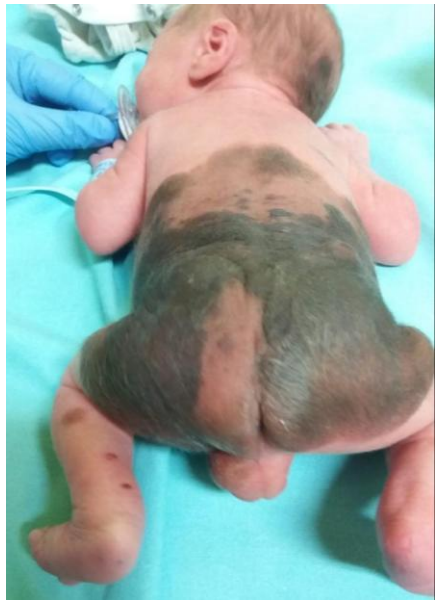
Treatment of AVM: symptomatic

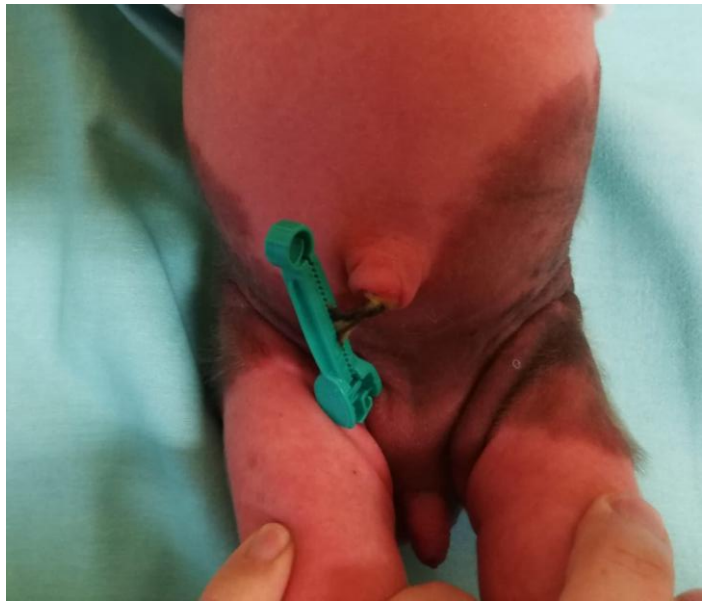
LORENZO

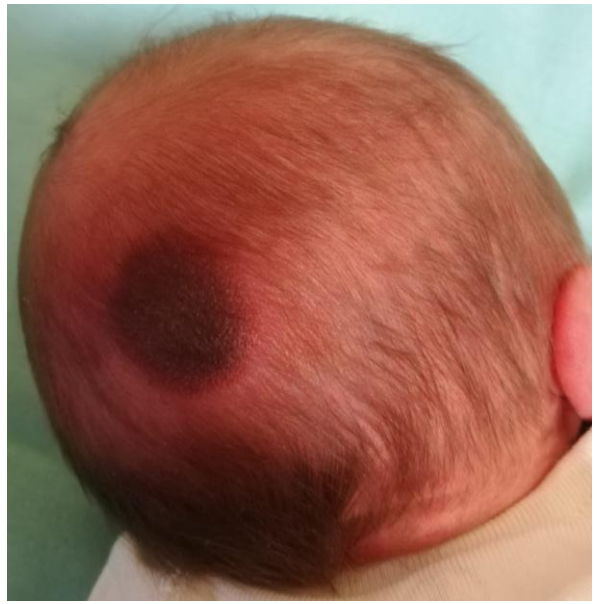
4-month-old boy born at term from non-consanguineous parents

At birth, he presented a bathing trunk giant congenital melanocytic nevus (GCMN) with multiple satellite nevi

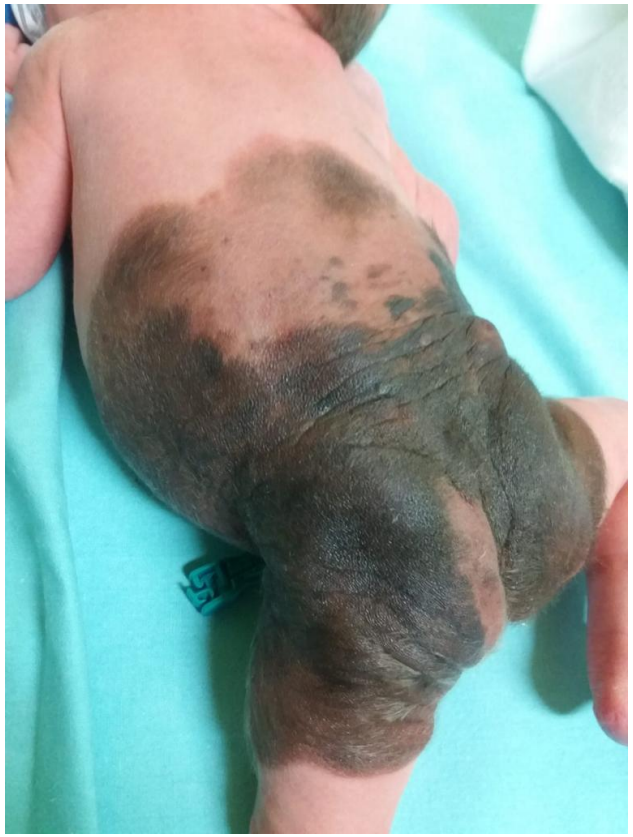
Brain and spinal MRI performed at 2 months of age to investigate a neuromelanosis did not show pathological findings

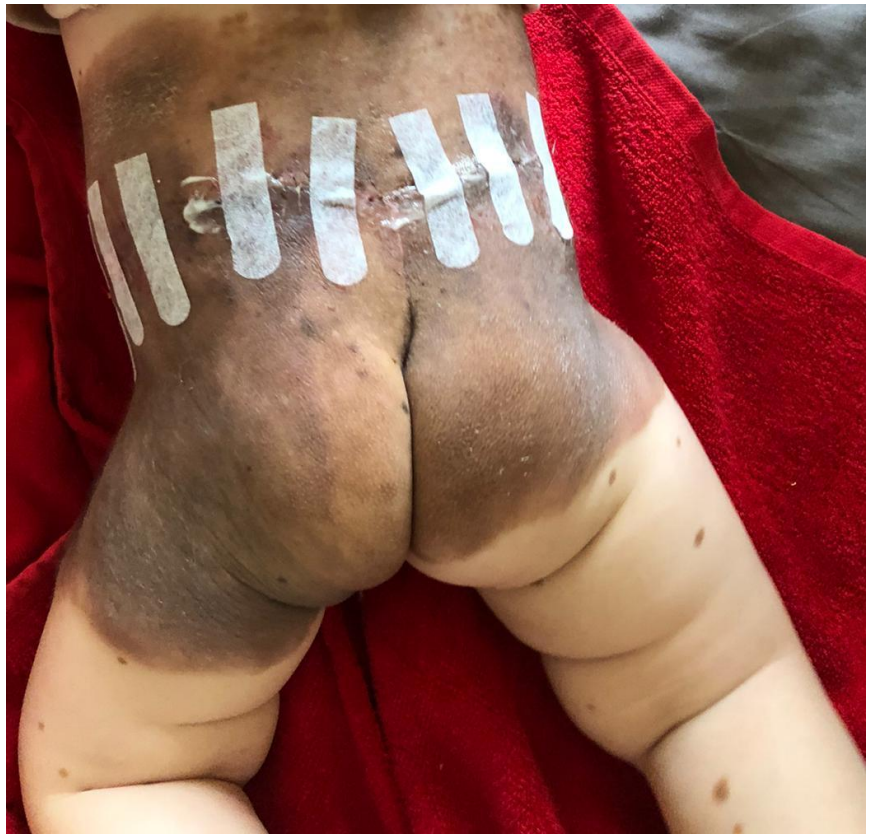




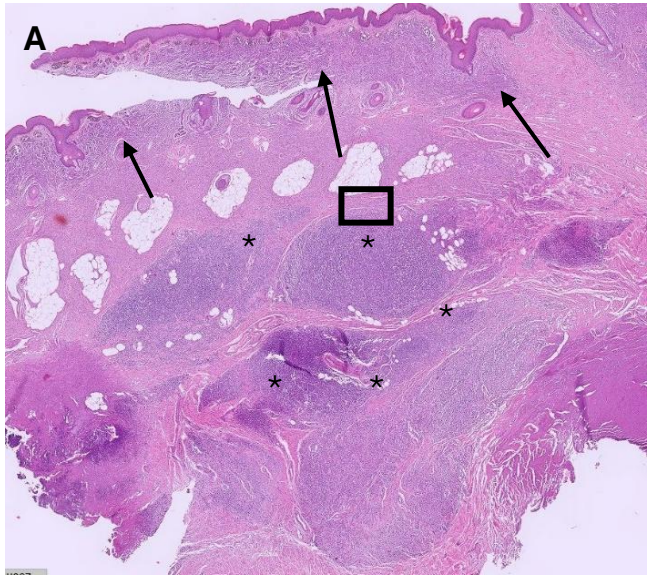


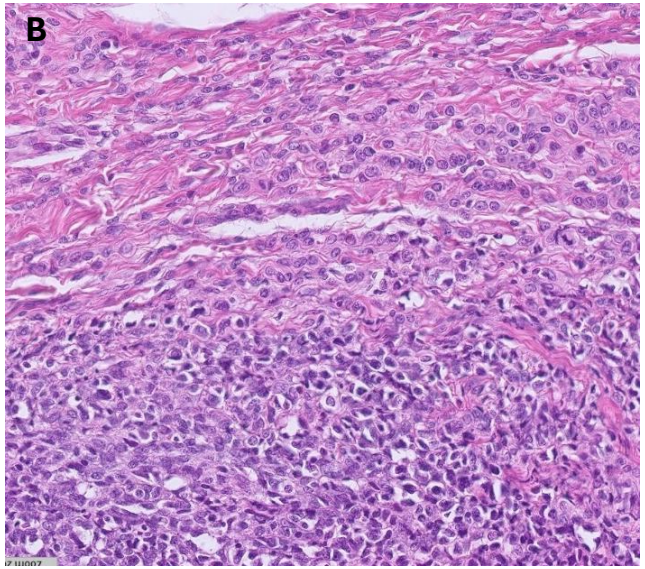
It was decided to treat him surgically with serial excisions, starting at 4 months of age
At histopathology, an unexpected polylobate, and poorly demarcated 0.9 cm sized nodule was detected in the hypodermis

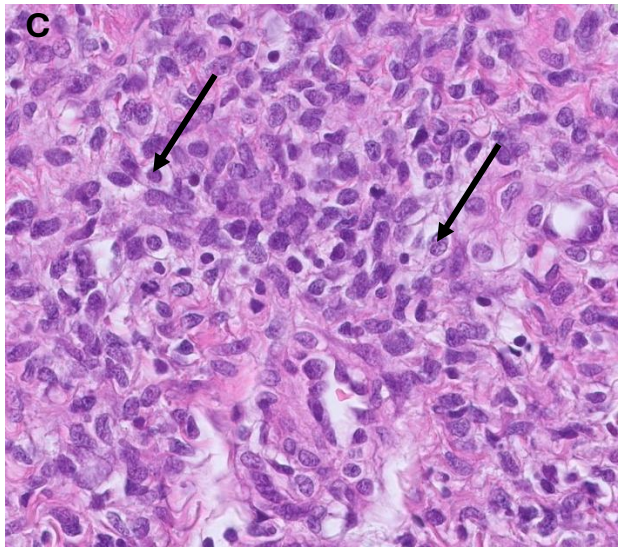




Histopathology





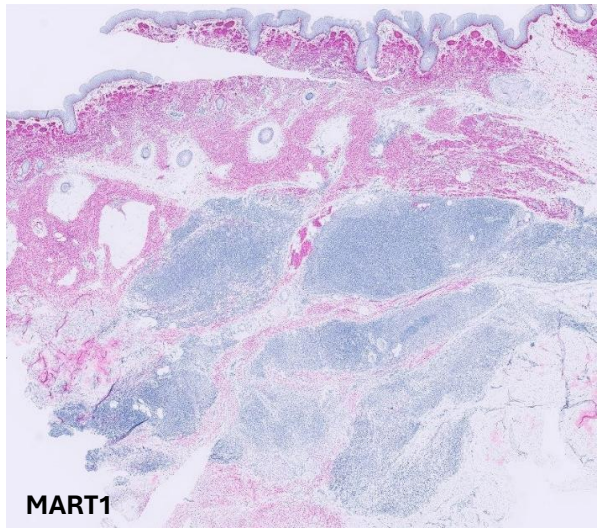


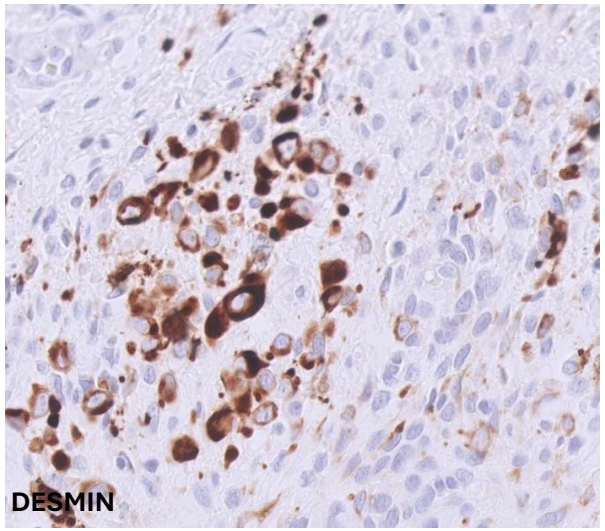
A. Low-power view of the congenital melanocytic nevus (arrows) and multiple nodules located in the hypodermis (stars)

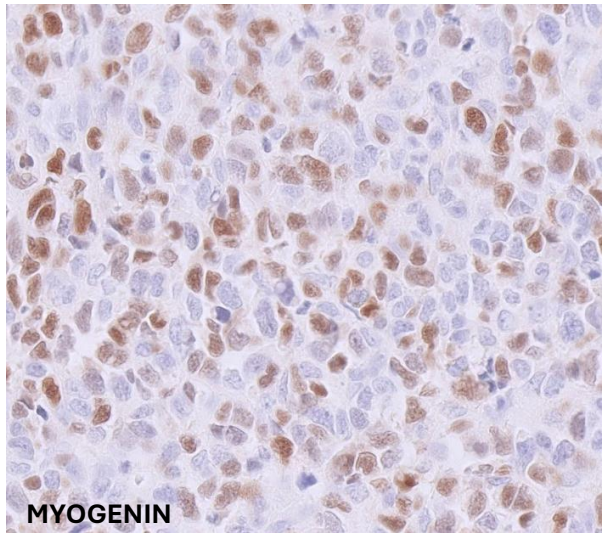
B. Medium-power view of the area comprised in the inset in A showing the interface between the nodular lesions and the background GCMN

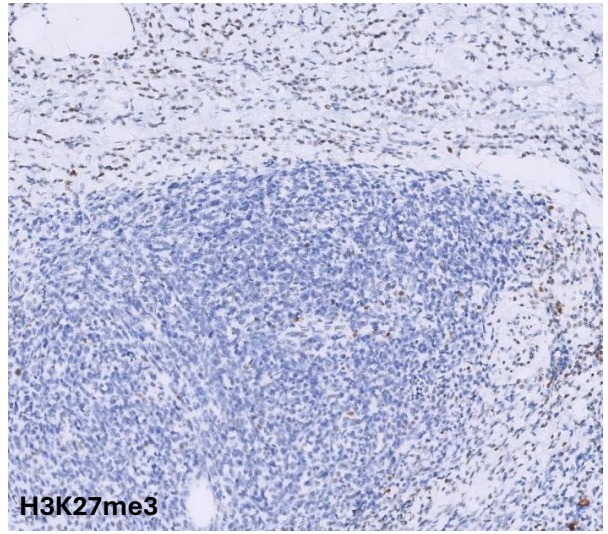
C. High-power view: the nodules were composed of medium-sized cells with round to ovoid hyperchromatic nuclei, a high nucleus-to-cytoplasm ratio and frequent mitoses (arrows)

Immunohistochemistry









MART1 immunostaining highlights the negative nodules in the positive GCMN background.

Focal expression of desmin and myogenin in the nodules, and MyoD1 focally expressed

Loss of H3K27me3 expression is lost observed in > 80% of malignant cells, while it is maintained in the GCMN

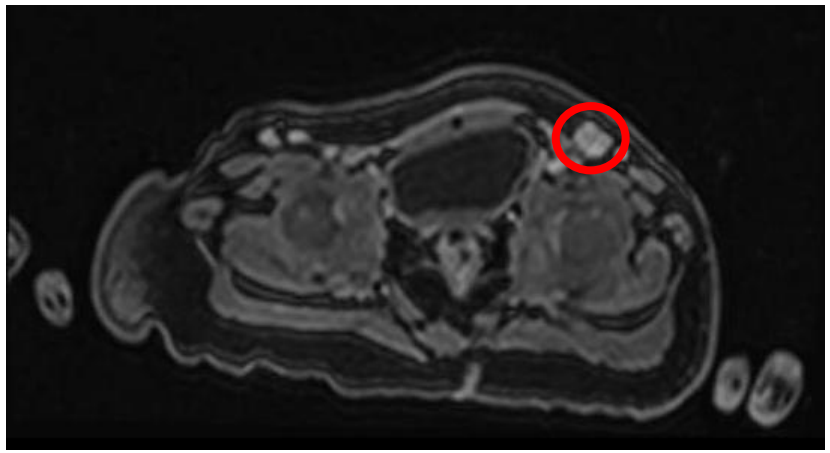
Diagnosis and Staging

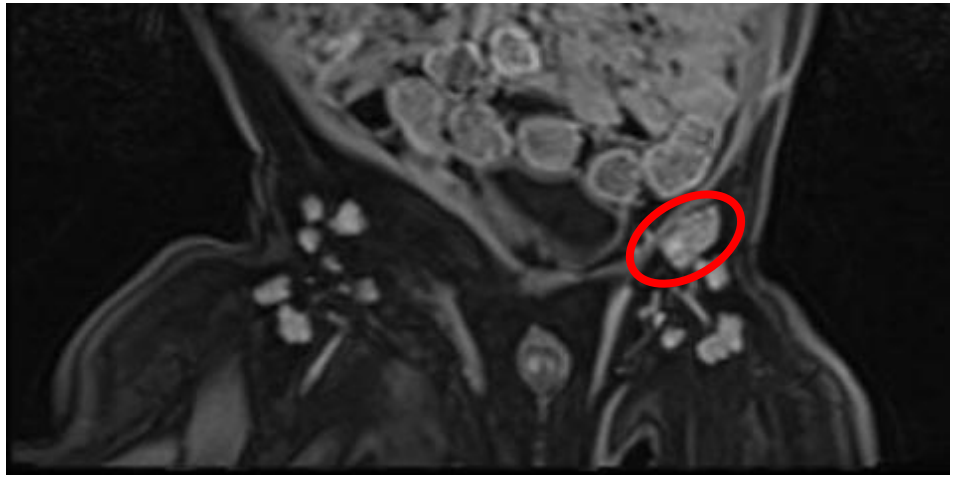
Altogether pathological and immunohistochemistry findings support the diagnosis of:

EMBRYONAL RHABDOMYOSARCOMA

(ERMS)

Staging exams showed on MRI a left inguinal lymph node involvement at 2 months from diagnosis





GCMN AND ERMS

GCMN are a rare subset of benign melanocytic tumors present at birth and defined as having an expected diameter in adulthood over 40 cm

The driver mutation usually involves NRAS (about 70% of GCMN), followed by BRAF (7%) genes

GCMN may give rise to several benign and malignant tumor types (e.g., melanoma, rhabdomyosarcoma, neurocristic hamartoma, plexiform neurofibroma, etc.)

RMS develops in rare GCMN, and may associate with melanoma, with 15 cases described up to now including 3 hybrid tumors

The large majority of reported RMS are ERMS, and occur in infants, with a median age at diagnosis of 14 months

Pitjadi TM et al Dermatopathology (Basel). 2019 Jun 26;6(2):91-98.

Treatment and prognosis

The patient was treated according to RMS chemotherapy protocols and he is in remission at 50 months from diagnosis, and in regular follow-up for the GCMN

Although the majority of patients developing RMS on GCMN reported in the literature (15) progressed or died within a median time of 11 months from diagnosis, a 1/3 of them remained free of disease at a median follow-up time of 40 months, like our patient