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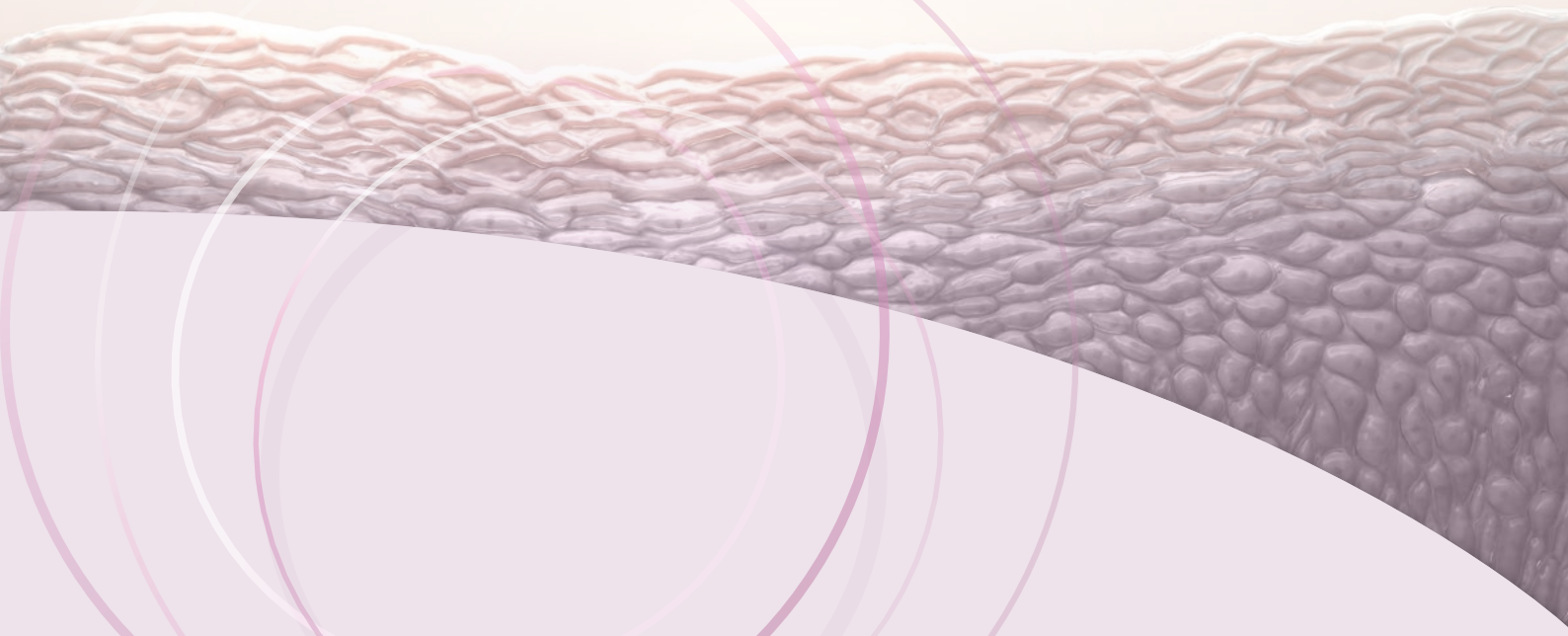
EUROPEAN ACADEMY OF
DERMATOLOGY AND VENEREOLOGY

Information Leaflet
for Patients



VASCULAR ANOMALIES IN INFANCY

The aim of this leaflet

This leaflet is designed to help you understand more about vascular anomalies in infancy (particularly the two most common forms: Capillary Malformations and Infantile Hemangiomas). It tells you what these conditions are, what causes them, and their diagnosis and treatment options.

VASCULAR ANOMALIES IN INFANCY

What are vascular anomalies in infancy?

Vascular anomalies are blood vessel abnormalities that represent a huge number of infantile disorders. Fortunately, the vast majority of these do not indicate a serious problem. However, the diagnosis should be confirmed by an expert in order to exclude symptomatic forms or *comorbidities* (other medical conditions) that are rare but still occur.

In the present classification adopted by the International Society for the Study of Vascular Anomalies (ISSVA) in 2014, vascular anomalies are divided into *vascular tumors* and *vascular malformations*. *Vascular tumors* may be benign, borderline, or malignant.

Vascular malformations may be simple (including *capillary malformations*) or combined. While mutations of many genes are responsible for a large proportion of *vascular malformations*, the origin of the most common vascular anomalies in infancy (i.e. *infantile hemangioma*) is still unknown.

CAPILLARY MALFORMATIONS

What are the main types of capillary malformations?

Capillary malformations (CM) are low blood flow vascular anomalies affecting the skin and mucosal layers. Two main subtypes of capillary malformations can be identified:

1) Medial congenital maculae

Synonyms: nevus flammeus neonatorum, nevus simplex, "salmon patch," or *"fading capillary stain"*

This is extremely common in Caucasian babies, clinically appearing as a birthmark of pinkish-red color which fades when pressure is applied. In most cases, it typically disappears within the first years of life. The most commonly affected anatomical sites are, in decreasing order (along the center of the body): the neck and *occipital* (back of the head) area ("Stork bite"), mid-forehead ("Angel Kiss"), and sacral region ("Butterfly Mark").

2) Lateral congenital maculae

Synonyms: port wine stain (PWS), nevus flammeus

This is rare in all ethnic groups, presenting as a pinkish-red birthmark of variable size with sharp edges, which typically fades when pressure is applied. *Macula* (spots) can occur on any part of the body with a tendency for the face, where it often has a *segmental* (patchy) distribution.

Extension to the *mucosal* surfaces (mucous membranes such as lips and in the mouth) is possible. The distribution of *capillary malformations* to the forehead can be the indicator of *Sturge-Weber syndrome*, where *leptomeningeal* (brain) and ocular (eye) involvement are observed. The distribution of *capillary malformation*s to a lower limb (i.e. leg) can be associated with venous and lymphatic abnormalities (*Klippel-Trenaunay syndrome*).

What do capillary malformations look like?

Capillary malformations are persistent flat birthmarks which vanish when pressure is applied, ranging in color from pink to red-purple. The affected skin has a normal temperature. In newborns, *capillary malformations* can sometimes be confused with *infantile hemangiomas* during its prodromal phase, but the misdiagnosis can be easily avoided with a simple evolutionary observation since capillary malformations are stable, while *infantile hemangiomas* evolve with a rapid proliferation. The dermatologist may also consider *arteriovenous malformations* (an abnormal connection between arteries and veins) in their early phase as another possible diagnosis.

What is their natural evolution?

While the color intensity of *medial maculae* ("salmon patch") fade within a few months of life, *lateral maculae* (port wine stain), may change over the years to a purple color. Sometimes the affected skin evolves towards a nodular *hypertrophy* (excessive growth) in adulthood with a typical thickening or "cobblestone" appearance. Furthermore, lateralized forms may be associated with *hypertrophy* of the affected area.

How are capillary malformations diagnosed?

The diagnosis of *capillary malformations* is essentially a clinical one. Color-Doppler ultrasound scan is the first study used for complex vascular malformations, followed by MRI (Magnetic Resonance Imaging).

How are capillary malformations treated?

Laser photocoagulation by vascular lasers is the first-choice treatment for *capillary malformations*, especially for the facial location. This treatment should preferably be started in childhood, and surgery should be considered only in selected cases.

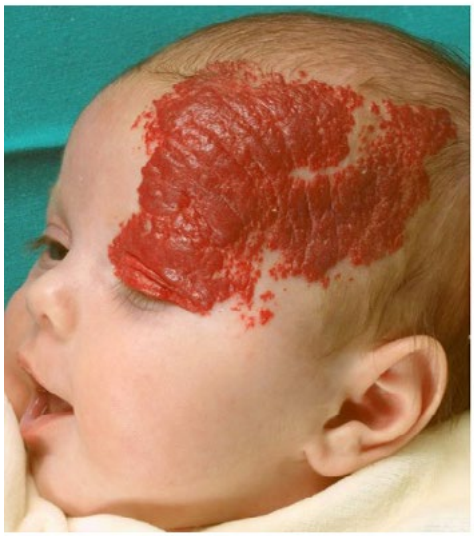


Fig. 1a A 2-month-old girl with an impressive, segmental IH non responsive to oral steroid

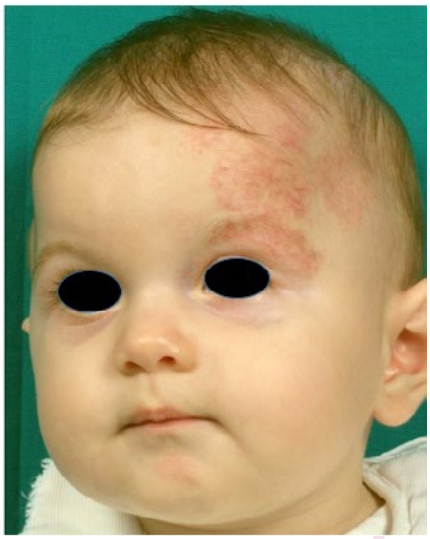


Fig.1b The same patient, at 6 months of age, after a short course of propranolol

INFANTILE HEMANGIOMAS

Who is affected by infantile hemangiomas?

Infantile hemangiomas (IH), or *hemangiomas* of infancy, are very frequent in newborns with an incidence of 3-10%. The cause seems to be multifactorial, but is still unknown. Female gender, Caucasian race, premature birth, advanced maternal age, *placenta previa*, and *preeclampsia* (conditions during pregnancy) are risk factors.

What do infantile hemangiomas look like?

IH more frequently appear in the first few days or weeks of life, often as a red, smooth, or *lobulated* (rounded), elastic nodule ("Superficial IH"). In "Deep IH," color is absent, and in "Mixed IH," color and swelling are both present. IH can appear in any part of the body, but head and neck locations are common. Although most IH are *non-syndromic* (not a pattern indicating a certain disorder), "Segmental IH" can be associated with underlying anomalies. Internal localizations are rare, but they must be investigated when there are multiple eruptive hemangiomas and/or *hepatomegaly* (liver enlargement) with signs of *congestive heart failure*. A liver ultrasound is recommended, as the liver is the most common involved organ (following the skin).

What is their natural evolution?

The IH life cycle is divided into 3 phases:

1. proliferating (rapid production) phase (usually 0-6 months)

2. resting phase (1-2 years)

3. involuting (shrinking) phase (2-10 years)

Photographs showing improvement of *Infantile Hemangioma*

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IH reach 80% of their volume in the first months of life. Only a minority of IH grows beyond the ninth month of life. The resting phase is characterized by a softening of the lesion and fading, starting from the central area with a progressive reduction of the volume and color. The involuting phase is characterized by a complete regression with possible residual secondary effects such as loose skin, *atrophy* (reduction in size), and/or *telangiectasias* ("spider veins") and/or *fibrosus residuum* (residual fat).

How are infantile hemangiomas diagnosed?

The diagnosis of IH is generally clinical. Some locations require a multidisciplinary approach (e.g. an ophthalmologist for *periorbital hemangiomas*; an otorhinolaryngologist (ENT) for laryngeal and auricular/ear involvement). In some cases, especially for "deep IH," the Color-Doppler ultrasound is helpful, while in rare cases a biopsy as well as MRI are necessary.

How are infantile hemangiomas treated?

Propranolol

Oral propranolol is the first choice of treatment for IH. Corticosteroids, vincristine, interferon (systemic) and timolol (topical) can be used for selected cases. Oral propranolol is indicated in the treatment of proliferating IH, such as life- or function-threatening IH, ulcerated IH with pain and/or lack of response to simple wound care measures, and IH with a risk of permanent scars or disfigurement. After an initial cardiology evaluation, treatment should be performed by clinicians with previous experience (in a clinical setting able to handle potential adverse reactions). Infants aged 5 weeks to 5 months can be treated, generally for a 6-month period.

Contraindications to propranolol treatment include asthma, low blood pressure, *peripheral vascular disease*, some heart diseases, and *pheochromocytoma* (type of tumor). Monthly monitoring at a hospital is recommended to check if there are side effects. In cases of abnormal sweating and irritability, a glucose test should be done. If there is vomiting, diarrhea, or lack of appetite, treatment should be postponed. In case of a low heart rate (<70 beats per minute [bpm], or <80 in newborns), heart problems, or a maternal history of *connective tissue disease*, an ECG and other cardiology evaluation should be carried out. Propranolol treatment does not need to be modified during vaccinations.

Laser

Laser *photocoagulation* is indicated as part of a multimodal treatment for ulcerated IH, deep facial IH, segmental IH of the mouth and upper respiratory tract, remaining *telangiectasias*, and results after the involuting phase. Lasers include the pulsed dye laser and neodymium: YAG laser, as well as recently a method that provides both using the same device.

Surgery

Indications for surgical treatment of IH are rare and similar to those of medical treatment (potentially life-threatening, risk of functional damage, cosmetic damage, or permanent ulcer). Surgery is the first-line treatment, in combination with or without laser treatment. Surgery should be done when medical therapy is contraindicated or when medical therapy fails or obtains only a partial effect, especially when the IH is localized in the facial region or around the mouth. It can also be performed when excess tissue remains af¬ter the involution phase. ■

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