

## Symposium: Dermatology

# Vascular birthmarks: update on presentation and management

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### Key points

- Vascular birthmarks are common in neonates and children.
- It is important to differentiate a haemangioma from a vascular malformation as management of these lesions is different.
- Most haemangiomas do not require treatment but those that do should be treated earlier rather than later. For difficult haemangiomas, referral to expert centres for further management should be considered.
- Children and families should have access to information from their general practitioners, paediatricians and community health staff.
- A multidisciplinary approach is mandatory for the complex paediatric problems associated with vascular birthmarks.
- For portwine stains, psychological and social support including advice on camouflage make-up should be available to help deal with the problems of disfigurement.

### INTRODUCTION

Vascular birthmarks are common in children with the incidence being one in three of all newborns. Although they are traditionally classified into two main groups as haemangiomas and vascular malformations,<sup>1,2</sup> the complexity and confusion with

classifying these lesions has necessitated the use of additional new terminology which is telangiectatic haemangiomas and telangiectatic malformations. Haemangiomas are dynamic, proliferative and endothelial anomalies with their hallmark being rapid growth; while vascular malformations such as portwine stain consists of dysplastic, ectatic vessels which persist throughout life. Telangiectases refer to superficial cutaneous vessels representing dilated venules, capillaries or arterioles which occur through the release or activation of vasoactive substances, e.g. anoxia, hormones, chemicals, infection and physical factors which result in vessel neogenesis. Some telangiectasias behave initially as haemangiomas and vascular malformations, hence the terminology, telangiectatic haemangiomas and telangiectatic malformations.

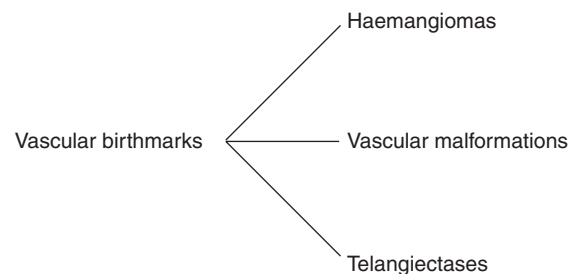


Fig. 1

### HAEMANGIOMAS

These are true endothelial tumours which in their proliferative phase have a high expression of proliferating cell nuclear antigen, vascular endothelial growth factor, type IV collagen, urokinase and basic

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**Fig. 2A** A 2-day-old baby with minimal evidence of a capillary haemangioma on the right side of the face. **B** The same baby as in Fig. 2A at 32 days of age with an ulcerated painful upper lip and the intense redness from the proliferation. **C** The same child as in Figs 2A & B at 1 year old. Note the massive haemangioma causing amblyopia in the right eye and the deformation and disfigurement caused by this lesion.

fibroblast growth factors. This suggests active angiogenesis and confirms haemangiomas to be completely different from vascular malformations.<sup>3</sup>

Haemangiomas are further sub-classified into the following main groups.

#### Classification of haemangiomas

1. Capillary/Strawberry naevus (red)
2. Cavernous (deep and bluish)
3. Mixed (superficial and deep)
4. Verrucous (warty and dark)
5. Multiple haemangiomas
6. Neonatal haemangiomatosis
7. Lymphangio-haemangiomas (blood lymph blisters)
8. Haemangiomas associated with syndromes
9. Haemangiomas associated with major blood vessel abnormalities.

#### Capillary haemangiomas

The most common is the strawberry naevus with an incidence of 1:20 babies. There is a preponderance in female and preterm neonates. Over 80% of these will regress spontaneously by the age of 7 years, however, a few will cause problems such as bleeding, ulceration, deformation and disfigurement (Figs 2A-C). For rapidly proliferating lesions that are at a site which will cause a problem, early treatment with oral or intra-lesional steroids, alpha-interferon and laser therapy should be considered. Occasionally, combination therapy should be instituted.

#### Capillary haemangiomas

- Usually not present at birth
- Appear within first few weeks of life
- Up to 3 months there is rapid proliferative growth
- Maximum size is usually reached by 6 months
- Static for up to a year, then slowly regress spontaneously
- Complete resolution by 5–7 years
- Residual telangiectatic lesions may persist.

#### Complications of capillary haemangiomas

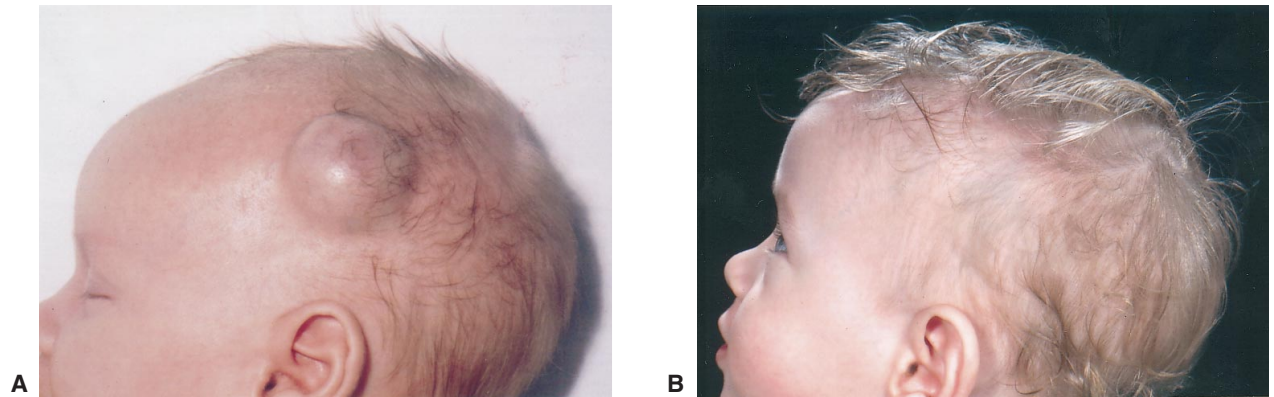
- Bleeding
- Ulceration/infection
- Deformation/disfigurement
- Impairment of vision
- Airways obstruction (subglottic)
- Major blood vessel abnormality
- Digital gangrene
- Anomalies of thorax and neck.

#### Cavernous haemangiomas

These are deep and bluish in colour. They are composed of possibly larger venules which are clustered together and located deeper into the skin and, hence, the blue colouration. Almost all will resolve naturally without treatment (Figs 3A & B).

#### Mixed haemangiomas

In these lesions there is a combination of superficial (red) and deep (bluish) vascular components. Most



**Fig. 3A** A 1-month-old baby with a cavernous haemangioma. **B** The same child as in Fig 3A with complete regression of the cavernous haemangioma by 1 year of age. The process occurred by natural resolution.

will disappear completely with time and no treatment is required but in some rapidly proliferating life-threatening lesions especially around the vital functional organs, e.g. eyes, nose, mouth and perineum treatment should be started urgently. This could be by compression bandage, steroids, or surgical intervention with embolisation or removal of the lesion depending upon the nature and site of the haemangioma. Alpha-interferon has also been used with variable response.

Rarely, a rapidly proliferating lesion may cause platelet consumption leading to thrombocytopenia and resulting in disseminated intracoagulopathy. This condition is known as Kasabach-Merritt syndrome.

#### **Verrucous haemangiomas**

These are uncommon congenital haemangiomas present from birth where there is unilateral hyperkeratotic lesions mostly seen on the lower extremities. Clinically, they are warty, crusty and dry dark lesions (Fig. 4). With age they can bleed and cause difficult management problems. Pulse dye laser therapy may stop the bleeding, flatten the lesion, reduce hyperkeratosis and may lessen the pain and discomfort.

#### **Multiple haemangiomas**

These are individual separate cutaneous capillary haemangiomas scattered all over the body. They sprout out at different stages in the first few weeks of life. Usually they do not cause any problems unless internal lesions are also present. Babies should be investigated at an early stage with an abdominal ultra-sound scan, a cranial CT scan and echo-cardiogram, if appropriate, to look for internal manifestations.

In blue rubber bleb naevus syndrome there is angiomas characterized by numerous cavernous like haemangiomas that involve the skin, mucous membrane and other parts of the body like gastrointestinal tract, lips, oral cavity, glans penis, nasopharynx and rarely brain meninges and heart.

#### **Neonatal haemangiomatosis**

This is a rare condition which can be life-threatening. There are many miliary blood-filled circular individual lesions not only in the skin but also internally. Within the first 4 weeks of life the baby may present with congestive cardiac failure, liver failure and may succumb to multi-system organ failure. Treatment with steroids should be started following the diagnosis of internal lesions and age of the patient. Alpha-interferon may also be considered as part of the treatment regime. However, each failing system should be treated individually.

#### **Lymphangio-haemangiomas**

These are a mixture of lymphatic and blood vessel abnormalities all amalgamated together (Fig. 5). They



**Fig. 4** Very extensive verrucous haemangioma.



**Fig. 5** Very extensive lymphangio-haemangioma. Note the lymph and blood blisters including the vericosities.



**Fig. 6** A 5-day-old baby with an extensive portwine stain on the face.

create difficult management problems because of the nature of the abnormalities involved.

#### Haemangiomas and syndromic associations

Numerous syndromes have been identified with vascular lesions.<sup>4</sup> Some examples are Beckwith-wiedemann, Trisomies 13 and 18 and Maffucci syndromes.

#### Haemangiomas associated with major blood vessel abnormalities

Capillary haemangiomas on the head and neck may have associated cardiovascular abnormalities, e.g. coarctation of aorta<sup>5,6</sup> or they may present with subglottic haemangiomas. Any infants who present with haemangiomas in these areas should be investigated to exclude cardiovascular and subglottic involvement. One should be aware of minimal respiratory distress in a baby which may be the presenting symptom of proliferating haemangioma in the subglottic region.

### VASCULAR MALFORMATIONS

These are developmental abnormalities of blood vessels which consist of dysplastic ectatic capillaries that tend to persist throughout life and grow proportionately with the child. These can be slow, fast flow or with arterio-venous shunting. They can rarely have complex combined anomalies and some have syndromic associations, e.g. Proteus syndrome, Klippel treunay syndrome.

### PORTWINE STAIN

The most common vascular malformation is a portwine stain with an incidence of 3:1000 births. A portwine stain is also known as naevus flammeus and is defined as a vascular malformation of developmental origin characterized pathologically by ectasia of superficial dermal capillaries and clinically by permanent macular erythema (Fig. 6). It is present from birth and is often present on the face. This type of birth mark becomes darker, thickens with age and forms progressive nodularity and blebbing on the maturing portwine stain often resulting in major disfigurement. Recent advances have shown that pulse dye laser therapy is the main stay of treatment. Our experience supports the view that younger children age 6 months – 4 years tend to have a better response to laser treatment than older children. The aim should be to complete treatment prior to starting main stream education so that psychological and social interactions are as normal as possible (Figs 7A & B).

Portwine stains can be associated with other medical problems. These are discussed as follows:

#### Glaucoma

A portwine stain around the eye may cause ophthalmic involvement leading to ocular problems especially glaucoma. The incidence of glaucoma varies between 15–25%. It is recommended to examine all children's eyes by an ophthalmologist as soon as possible and then on a regular basis thereafter.



**Fig. 7A** A baby with a portwine stain aged 4 months. **7B** The same child at 1 year and 9 months after four laser treatments.

### Sturge-Weber syndrome

There is an association of a facial portwine stain with angiomas and calcification in the leptomeninges over the cortex. The individual presents with convulsions, hemiplegia/hemiparesis and mental retardation. There may be associated visual disturbances. The risk of a unilateral facial portwine stain having an associated abnormality of the Sturge-Weber syndrome in our series at Great Ormond Street Hospital was 25% and for bilateral portwine stain was 75%. All children with facial portwine stains on the upper part of their face and scalp should have a baseline MRI scan with gadolinium enhancement performed to exclude central nervous system involvement.

Facial portwine stains on the lips commonly cause soft tissue hypertrophy leading to unsightly enlargement of the lips. Early referral to plastic surgeons is recommended for possible debulking procedure of the disfigured lips.

### Klippel Trenaunay syndrome

An extensive vascular malformation of any limb may cause soft tissue and bony hypertrophy (Fig. 8) leading to an overgrowth of that limb which can become enormous and can be associated with severe varicosities, thrombotic episodes, thrombophlebitis, venous

stasis, ulceration and recurrent infections. Occasionally, portwine stains may be extensive so as to cover very large areas of the skin.

### Klippel Trenaunay Weber Syndrome

This condition was described by Parkes Weber in 1918<sup>7</sup> with clinical features of a portwine stain on a limb present from birth, enlargement of that limb and presence of arterio-venous anastomoses. Less commonly, there is also involvement of lymphatics and more complex blood vessel anomalies.

Both of the above conditions require multidisciplinary management (Fig. 9). Due to the significant degree of variations in these conditions, medical care should be tailored to the individual's needs.

### Proteus syndrome

This was first described in 1983 by Wiedemann et al.<sup>8</sup> although cases have been written up dating back to 1856. In this congenital condition the abnormalities are present from birth although these may only become apparent and develop with age. There is overgrowth and enlargement of soft tissue and bone affecting any area of the body. Most often it involves hands and feet, especially one or more fingers or toes.



**Fig. 8** A 7-year-old child with extensive vascular malformation and soft tissue hypertrophy of the right leg. Note the discrepancy in length and width between the two legs.

The skull may also be involved and sometimes there is hemi-hypertrophy of one side of the body. There is the presence of vascular malformations and there may be other cutaneous involvement like epidermal naevi and soft but deeper subcutaneous lumps. Bony problems may involve the spine.

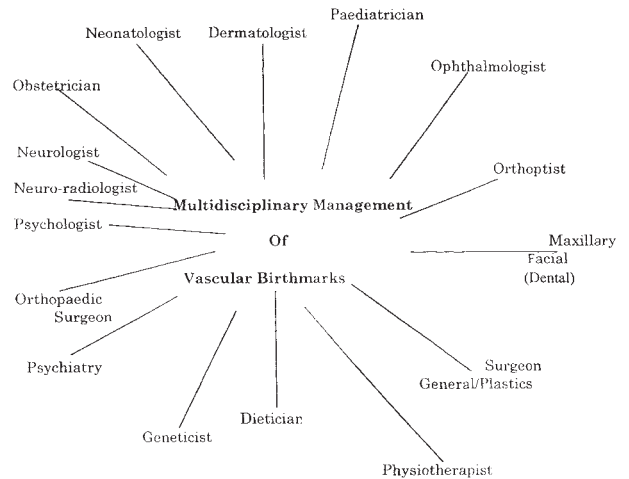
The presentation of the abnormalities is variable from minimal complications to severe involvement depending on the nature of the problem, the site and the severity. Coordinated multidisciplinary specialist care is required to cater for these individuals. Investigations should depend on individual problems and these may vary from baseline blood tests to full radiological assessment including CT or MRI scans.

**TELANGIECTASES**

Telangiectases are malformations of the superficial cutaneous vessels measuring 0.1–1.0 mm and represent dilated capillary, venous and arteriole vessels. These are classified into telangiectatic haemangiomas and telangiectatic vascular malformations.

**Telangiectatic haemangiomas**

These lesions may or may not be present from birth but they present as spiral or papular telangiectatic



**Fig. 9** Many disciplines can be involved in the care of a child with a vascular birthmark.



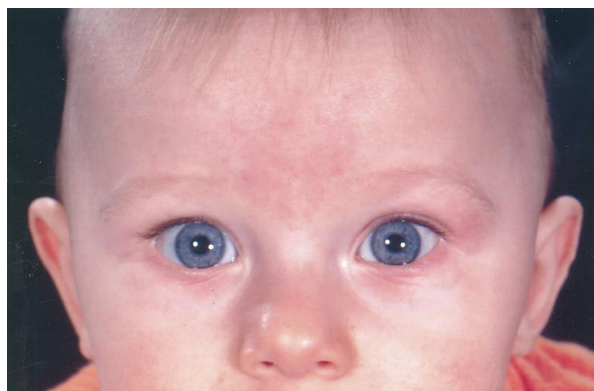
**Fig. 10** A 4-month-old baby with a bilateral telangiectatic haemangioma on the buttocks. Note the ulcerations and the sparing of the anal region.

lesions which proliferate very slowly. There is absence of a rapid growth phase. Most present bilaterally and symmetrically especially on the face and on the perineum. They often have other associated medical problems. There is a tendency to bleed and ulcerate just like the capillary haemangiomas (Fig. 10).

**Telangiectatic vascular malformations**

The most common telangiectatic vascular malformation is a salmon patch also known as a stork mark with an incidence of 1:40 babies. This is usually present at birth but disappears by the age of 2 years. The most common site is the forehead and the eyelids (Fig. 11). The telangiectatic lesion on the nape of the neck usually persist throughout life.

The next most common is the spider naevus which is a common presentation in young children. Other forms are spiral, linear, papular, multiple and matt telangiectasias. All respond extremely well to pulse dye laser therapy with an excellent outcome.



**Fig. 11** An 8-month-old child with a salmon patch.

### **Cutis marmorata telangiectatica congenita**

This is a rare condition present from birth and characterized by a distinctive reticulate pattern which presents as a livid, reticulate mottling of the skin. It mostly involves circumscribed segments of the skin, usually the limb, but can be widespread (Fig. 12). This should be distinguished from physiological cutis marmorata which apparently presents at birth but only lasts for less than a week after birth. Cutis marmorata telangiectatica congenita is darker, sharply delineated, segmental and may be associated with other medical problems, for example, phlebectasis, ulceration with crusts, mild skin atrophy, verrucous or hyperkeratotic lesions.

### **Telangiectases with syndromic associations**

The commonly known association are the dominant conditions, essential telangiectasia and Ataxia telangiectasia; others of note are Rothmund Thomson syndrome, Osler-Weber-Rendu syndrome and Goltz syndrome.

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**Fig. 12** A 5-year-old girl with extensive Cutis marmorata telangiectatica congenita.

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