

A CORE SURGICAL TRAINEE'S GUIDE TO: VASCULAR MALFORMATIONS IN CHILDREN

J Davidson & K Ford

A Core Surgical Trainee's Guide to: Vascular Malformations in Children Pediatric Surgery

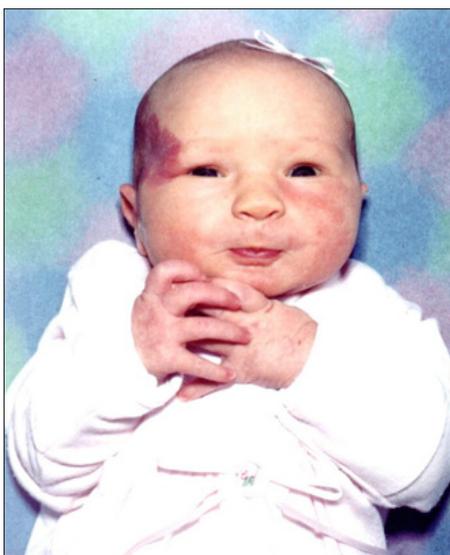
Abstract

Vascular malformations can prove to be a daunting group of conditions to the surgical trainee. Historically, difficulties in classification and nomenclature, along with a lack of a definitive responsible specialty, have led to variability in management strategies. Here we hope to give a brief overview of vascular malformations, detailing the four most common a little further.

Keywords: *Vascular malformations, paediatric.*

Case Example

A mother brings her 3-week old baby girl to see you in the paediatric surgical clinic, having been referred by the GP a week ago. Her baby was born at term in good health. From birth, she was noted to have a purplish discolouration of the upper portion of one side of her face. A diagnosis of a capillary malformation (Port Wine stain) being made, she wishes to know the available treatment options, whether this lesion will improve with time, and if it carries any potential serious associated consequences.



Background

Vascular malformations can be defined as an abnormal structuring of histologically normal vascular tissue (1). As a group of conditions, vascular malformations have an incidence of approximately 1.5% of live births (2).

Like all congenital anomalies, vascular malformations are always present at birth, although they may not be detected for the first few months of life. They display a normal rate of endothelial growth and enlarge by hypertrophy rather than proliferation, growing at the same rate as the child. Unlike haemangiomas, vascular malformations do not involute.

Vascular malformations are not thought to display a pattern of inheritance, rather to be caused by sporadic embryonic mutations in the genes responsible for regulating endothelial development (3). There are certain associated genetic and chromosomal abnormalities: lymphatic malformations are associated with a number of conditions, including trisomies 21, 13 and 18, there is also the hereditary telangiectasia syndrome (Osler-Weber-Rendu) where arteriovenous malformations may present at a number of anatomical sites.

Nomenclature has historically been difficult to appreciate on account of eponymous siring of different lesions, however the anatomical classification of Mulliken and Glowacki renders the classification of vascular malformations more palatable to the journeyman trainee (4). Within the classification, malformations can be grouped into low and high flow (those involving an arterial component).

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Classification	Previous terminology	
<i>Single malformation:</i>	Capillary	Port-wine stain
	Arterial	
	Venous	
	Lymphatic	Cystic hygroma, Lymphangioma
<i>Combined malformation:</i>	Arteriovenous (AVM)	
	Capillary-venous-lymphatic	Klippel-Trénaunay
	Capillary-arteriovenous	Parkes Weber
	Combined	Sturge-Weber

Presentation

Presentation of vascular malformations is highly variable; however careful history and thorough physical examination is sufficient to establish a clinical diagnosis in most cases. Symptoms may be absent altogether or life threatening if it is an AVM. Even simple capillary malformations can be a source of severe cosmetic distress to a child. It is often advisable to use photography both as documentation of progression or resolution of the lesion, which may inadvertently act as a reassurance tool for the anxious parent.

Large vascular malformations are associated with a soft tissue mass visible on plain radiograph, sometimes with associated bony changes (5). Skeletal hypertrophy and shape alteration are more commonly seen with low-flow lesions, while reduced bone density is more common in high-flow malformations. Further diagnostic imaging including CT, MR and angiography can all provide valuable information in terms of classification and direction of therapy.

Management

Management of vascular malformations can loosely be broken into conservative, interventional and surgical (6). Owing to the rarity and complexity of this group of conditions, it is generally accepted that they be managed in large tertiary centres with a fully functional multi-disciplinary team at hand.

Interventional therapies range from the use of phototherapy, to embolisation, to the use of sclerosant agents. Surgical excision is achievable but, as later explained, often challenging and requiring the input of several surgical specialties. The multi-disciplinary approach is key in terms of assistive therapy in cases of large debilitating lesions, and psychology input may be desirable owing to the disfigurement that can develop from a young age (7).

Types of Vascular Malformation

Capillary Malformation

Capillary malformation, CM, may be limited or extensive, and present as a purplish discolouration of the skin (hence the now out-dated term, "Port Wine Stain".) Their pathophysiology is poorly understood. These lesions tend to darken and develop nodularity as the child grows older (8). 45% of CM are restricted to one region of the trigeminal dermatome, however they may occur anywhere on the body, crossing dermatomes and indeed the midline.



Figure 1: infant with CM of the left side of face, representative of Sturge-Weber syndrome.

Lesions of the upper face (i.e. ophthalmic branch of the trigeminal nerve) should prompt a consideration of a diagnosis of Sturge-Weber syndrome (SWS) (Figure 1), where associated ocular and leptomeningeal vascular malformations can lead to seizures, impact upon motor and cognitive function and retinal function.

Whenever a capillary malformation affects the forehead or scalp (as in the case example), particularly if the lesions are bilateral, the child should be referred for a contrast (gadolinium) MRI scan to rule out SWS. If SWS is diagnosed, the child's ongoing care would be shared between the paediatricians and neurologists. If there is no involvement of the brain or eyes and SWS has been ruled out, the capillary malformation becomes a cosmetic consideration only.

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CM can also be associated with developmental defects of the neural tube. These should be suspected in any lesion overlying the occiput or spine.

The treatment is cosmetic: laser photocoagulation demonstrates some improvement in 80% however as many as 50% notice some darkening of the lesion in the subsequent few years.

Lymphatic Malformation

Lymphatic malformations, LM, can be further divided (with the help of radiology and histology) into microcystic, macrocystic or combined (6). These present as a soft swelling, commonly of the head and neck, axilla and chest or buttocks and perineum. Overlying skin is usually normal but may be hold a bluish tinge on closer inspection.

Indications for ablative or excisional therapy include recurrent complications with infection, cosmesis, deformity, dysfunction, and leakage into body cavities or from the skin (3,8).

Surgical resection provides the only means of a definitive cure, however, this is largely only plausible for lesions that are well localized. Focal, macrocystic lesions are amenable to both sclerotherapy and resection. In contrast, more diffuse and predominantly microcystic LMs are difficult to eradicate by any method.

Head and neck LMs will often require maxillofacial input procedures to improve speech and swallowing issues related to bony overgrowth. Tracheostomy may be needed in cases of potential airway obstruction. When considered necessary, tracheostomy should precede any attempts at sclerotherapy, as reactive swelling can be dramatic in the initial period after sclerotherapy.



Figure 2: Facial lymphatic malformation.

Venous Malformations

Venous malformations, VM, represent the most commonly occurring vascular malformation. Examination will reveal a blue, soft and compressible mass. VM are often solitary but can be more extensive (2). Indications for treatment include: cosmetic, pain, reduced function, and significant bleeding. Much like LM, often all but the most simple, and consequently least problematic lesions are amenable to curative treatment.

For extensive VMs of the limbs, graded compression stockings can achieve satisfactory symptomatic relief. Intralesional sclerotherapy is the mainstay of treatment for most VMs, however cure with sclerotherapy alone is rare, due to a tendency for VMs to recanalise and enlarge after treatment. This propensity has led to a shift in expectation of treatment to controlling pain and impact on quality of life.



Figure 3: Venous malformation of the left cheek and lips.

Surgical resection is typically reserved for well-localized lesions but is complicated by morbidity and recurrence. Preoperative sclerotherapy is often utilised to shrink the lesion and decrease bleeding during the resection.

Arteriovenous Malformation

Arteriovenous malformations (AVMs) are most commonly sited within the skull (8). However these lesions can occur anywhere on the body. These 'fast-flow' lesions are usually noted at birth but are often misdiagnosed as capillary malformations or haemangiomas. The natural history of the fast-flow lesions is that of increasing erythema of the overlying skin and a palpable thrill developing. They have a propensity to enlarge rapidly with hormonal changes during puberty.

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Diagnosis can be confirmed with Doppler ultrasound which demonstrates feeding and draining vessels and arteriovenous shunting. Symptoms and complications of AVMs are due to the shunting that occurs within the lesion. AVMs also have potential to bleed or ulcerate. The Schobinger clinical staging system proposed is commonly used:

Stage I (quiescence)	Pink-bluish stain, warmth, and arteriovenous shunting evident on Doppler examination
Stage II (expansion)	As with stage I, plus enlargement, pulsations, thrill, bruit, and tense/tortuous veins
Stage III	As with stage II, plus dystrophic skin changes, ulceration, tissue necrosis, bleeding and pain
Stage IV	As with stage III with associated cardiac failure (decompensation)

Figure 4: Arteriovenous malformation of the medial aspect of right hind foot.

The mainstays of management are embolization, sclerotherapy, surgical resection, and reconstruction. Embolization or sclerotherapy may be used to control symptoms, however the effects are often temporary due to the growth of new vessels within the lesion. Often complete resection is not possible, or would result in severe disfigurement, particularly in a young patient.

Conclusion

Vascular malformations are a group of conditions with considerable associated morbidity. These children are best managed in expert centres with relevant experience. Interventional medical and surgical options exist, but these may be inadvisable for more complex lesions, and do not carry a guaranteed cure rate even for the most simple and limited. Camouflage techniques should be introduced alongside child psychology support; many of these lesions can be markedly disfiguring and carry considerable psychological burden.

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MCQs

1. Which are the most commonly occurring?

- Arterio-venous Malformation
- Lymphatic Malformation
- Capillary Malformation
- Venous Malformation
- Combined multiple (e.g. Sturge Weber syndrome)

2. Which of the following injections is not used in the treatment of venous malformation?

- Bleomycin
- OK-432
- Propranolol
- Ethanol
- 5-fluorouracil

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3. Which of the following syndromes has a recognised link to vascular anomalies?

- a. Russell-Silver Syndrome
- b. Beckwith-Wiedemann Syndrome
- c. Noonan Syndrome
- d. Prader-Willi Syndrome
- e. Angelman Syndrome

4. A macrocystic extensive lymphatic malformation affecting the neck, formerly named...

- a. Cyst of Morgagni
- b. Branchial cyst
- c. Thyroglossal cyst
- d. Cystic hygroma
- e. Meckel's cyst

5. Which of the following is the most appropriate advice to give to the mother in the introductory case example?

- a. This lesion is benign, it may enlarge over the next year but should resolve without intervention
- b. This lesion is unlikely to spread, it may become more evident as the girl grows. There is a small proportion associated with more severe vascular lesions.
- c. This lesion has malignant potential and the child should be immediately referred to paediatric oncology services
- d. This lesion is indicative of a severe life-limiting chromosomal abnormality and the child should undergo chromosomal analysis
- e. This lesion is likely to spread over the face of the child and early surgical intervention is advisable

Answers

1. d 2. c 3. c 4. d 5. b

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