# Vascular Anomalies

*Introduction*
This area of surgery has been clouded by a variety of confusing and often nonsensical classification systems. Only a brief knowledge of these conditions is required as appropriate referral, especially where the potential for overgrowth exists, as this may prevent serious deformity in these unfortunate patients. Terms such as cavernous or capillary haemangoma, port-wine stains as well as any references to a fruit, such as a cherry or a strawberry should obviously be discouraged!

*Overview and Nomenclature*
The current standard classification was described by Mulliken & Glowacki in *PRS*, 1982. It is called the **BIOLOGICAL CLASSIFICATION**, and takes into account the physical findings, natural history and cellular features. It divides anomalies of the lympho-vascular system into two broad groups: **tumours** and **malformations**.

## VASCULAR ANOMALIES

- **VASCULAR TUMOURS** *(Endothelial neoplasms)*
    - Haemangioma
    - Haemangioendothelioma (KHE – Kasabach-Merritt Phenomenon)
    - Haemangiopericytoma
    - Tufted Angioma
    - Angiosarcoma

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    - Pyogenic Granuloma *(Acquired inflammatory vascular "tumour")*

Not really a tumour !

- **VASCULAR MALFORMATIONS** *(Abnormalities of embryogenesis / maturation)*
    - Single Vessel Forms (Or based on dominant type)
        - Capillary Malformations (formerly called Port-wine stains)
            - Sturge-Weber Syndrome
            - Cutis Marmorata Telangiectasia Congenita (CMTC)
        - Lymphatic Malformations
            - Microcystic (formerly called Lymphangioma)
            - Macrocystic (formerly called Cystic Hygroma)
        - Venous Malformations

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        - Arteriovenous Malformations (AVM)

This is the only high flow lesion

    - Complex Forms (Just for completeness sake…)
        - Low flow Capillary-Lymphatico-Venous Malformation (CLVM) also known as "**Klippel-Trénaunay Syndrome**"
        - High flow Capillary Arteriovenous Malformation (CAVM) also known as "**Parkes-Weber Syndrome**"

# A. VASCULAR TUMOURS

Vascular tumours are neoplasms characterized by increased endothelial cell mitotic rates. The terms acquired and congenital should be used cautiously with vascular anomalies, as presence at a cellular level may not be clinically detectable until later in post natal life. The most important tumour is the haemangioma. Most importantly, the much touted Kasabach-Merrit phenomenon of thrombocytopenia does NOT occur with haemangomas, but only with the more uncommon and prognostically worse haemangioendothelioma.

## 1. Haemangioma

*Definition*

This is a true **vascular neoplasm** with a very **unique natural history**: rapid growth, slow regression and no recurrence. This is reflected in the 3 stages of a haemangioma: the proliferating phase, the involuting phase and the involuted phase.

*Incidence*

These are the most common tumours of infancy, up to **1:10** caucasians. **M:F = 1:3 to 5**. The incidence is significantly raised in **premature infants**. They are much less common in darker skinned patients. 80% are solitary, 20% are multiple. Craniofacial haemangiomas may be associated with airway (sub-glottic) involvement – take care during intubation!

*Clinical Features*

Haemangiomas manifest (usually) **within 2 weeks** of birth. A **herald spot** may be present, which could be a telangiectatic, red or purpuric macule. The clinical course is unique and should be discussed with the parents fully.

In the first year of life, the tumour undergoes **rapid growth** but rate does not predict ultimate size. **Superficial lesions** are raised, firm and a deep crimson colour. **Deeper lesion** are slightly raised, blueish or telangiectatic and warm.

Between 1 and 5 years the growth slows down and then begins to regress. Signs of regression include a **colour change** from red to purple, **central paleness** and greyish discolouration. The rate of regression has no correlation to depth, size, site, gender or colour. Involution is complete by **5 years in 50%** and **70% by 7 years**.

**Normal** appearance is restored **in 50% of** cases. Residual **telangiectasia or skin laxity** (due to elastic fibre destruction) occur, with **yellow patches or atrophic scarring**. **Alopecia** of the overlying scalp may occur. Hypertrophy is rare.

*Diagnosis*

**Clinical** diagnosis is often sufficient. **Ultrasound and MRI** are useful for visceral and intracranial disease exclusion, and **Tc-99 red cell** scans can also be used. **Biopsy** is a last resort as profuse bleeding may occur.

*Treatment*

Most haemangiomas are small, harmless tumours which can be **expected to regress** without further treatment. Parental reassurance is required and serial photography is useful for follow up. There are several instances where intervention before regression is indicated:

1. **Ulceration**, with pain, infection and bleeding, **not responsive** to simple dressings
2. Repeated and profuse **bleeding**, often associated with minor trauma
3. High output **CCF**
4. **Airway** or upper digestive obstruction
5. **Vision** impairing (deprivational or astigmatic ambylopia)
6. **Aural** impairing (deprivational mono-aural hearing)
7. **Speech** impairing
8. **Social** impairment (school going age)
9. Anticipated scar from excision better than scar from expectant management

*Treatment Modalities*

**Intralesional Steroid Injections**: Depo-medrol 3-5mg/kg/dose injected at low pressure via a tuberculin syringe (30G). 3-5 repeats are required at 6 to 8 week intervals. Can be used for ulcerative lesions. This is my preferred treatment option.

**Systemic Steroids**: Prednisone, 2-3 mg/kg/day is given orally for 2 weeks. IV can be used for subglottic stenosis, or GIT bleeds. The dose is tapered every 2 weeks. Live viral vaccinations should be withheld during this time. High dose steroids are anecdotally effective, but not supported by research. Cushingoid facies is expected. Reflux and infections are treated as required.

**Interferon-Alpha**: This is used as a second line therapy in cases of steroid resistance as the complications are more significant. It is anti-angiogenic and reduces β-FGF levels. Empiric dosing is 2-3 million units/$m^2$, SC daily. 6 to 12 months of treatment are required. Low grade fever is common. Neutropenia and hepatic dysfunction also occur. Spastic diplegia is the most serious side effect.

**Chemotherapy**: Vincristine and cyclophosphamide have been used for life-threatening haemangiomas. I have occasionally used low dose bleomycin (1-5 Units per session) for intra-lesional injection in resistant cases – caution should be exercised and skin necrosis may be encountered. For injection in the face, injection under digital subtraction angiography guidance is suggested. It is easier to obtain in private practice and this is what I usually do, despite the significant cost implication.

**Radiation**: Low dose radiotherapy was given in the past, but is no longer recommended.

**Embolization**: This is mainly used for visceral (hepatic) haemangiomas that are responsible for severe CCF. Improvement is transient (a bridge while awaiting an effect of another modality).

**Laser**: The usefulness of flash-lamp dye lasers is limited to cosmetic improvement of the superficial portion of a cutaneous lesion. Subglottic lesions can be excised with $CO_2$ laser.

**Surgical Excision**: This should follow the principles of maximal tissue preservation and concealed scar placement. Revision of the scars is done at a later stage. Refinement of the involuted phase lesions requires cosmetic surgery.

## B. VASCULAR MALFORMATIONS

Vascular malformations result from **errors of embryonic development**. The classification of these anomalies is either haematic or lymphatic in nature and also on the rheology, as slow-flow (ie, capillary, lymphatic, or venous) or fast-flow (ie, arterial), or as combinations of the above.

### 1.1 Capillary Malformations (CM)

*Clinical Findings*

CM can present anywhere. They are usually **solitary** and following a **dermatomal** distribution. Initially **pink**, they tend to **darken over time** and may become **nodular. Overgrowth** of the affected part occurs, but the degree of overgrowth tends to be mild and not usually clinically significant.

The syndromic variant of the usual CM is called Sturge-Weber Syndrome. This is the association of a facial **CM (trigeminal V1**, possibly V2/3) with **leptomeningeal vascular anomalies.** The patient may present with:

- **Meningeal involvement**
    - refractory epilepsy
    - hemiplegia
    - developmental delay
- **Occular involvement**
    - retinal detachment
    - glaucoma
    - blindness

Plastic Surgery, neurosurgery and ophthalmology assessments are required. MRI is better than CT.

*Treatment*

This is often challenging.

- Cosmetic cover-up (**make-up**)
- Laser - **585nm Pulsed dye laser** helps with lightening, but penetration is 1.2mm and recurrence is common, but excellent results are being reported by some centres. Laser is best done at a young age, most centres aim to start at about 1 year old. It is very painful and must be done under GA.
- Surgical Excision - small lesions are easily **excised**. Excision and **resurfacing** with graft, flap or tissue expansion can be used. **Orthognathic surgery** for occlusal cants.

## 1.2 Lymphatic Malformations (LM)

These malformation may occur as microcystic or macrocystic or mixed forms. Older terms such as lymphangioma and cystic hygroma are confusing and should be discarded (along with the fruit!).

*Clinical Findings*

The malformations may be present at birth or may become evident within 2 years. LM are **usually solitary**, and can occur anywhere. The overlying **skin is normal** with a **blueish hue** (due to cystic fluid fill spaces). **Bleeding into these cysts** change them to dark red. Facial **overgrowth is common**, and may be severe. MRI is the best imaging medium.

*Treatment*

This is aimed at preventing complications and improving function. **Antibiotics** are only indicated for overlying cellulitis. **Bleomycin sclerotherapy** is widely used and can be quite successful. Surgery is tedious and has a very high complication rate. The operative goal is **complete resection**, as re-operation is even worse.

## 1.3 Venous Malformations (VM)

These are the most common of the low-flow vascular malformations.

*Clinical Findings*

They are usually **not apparent at birth**, but present during childhood. They are blue, soft and easily compressable. Most VM are sporadic and solitary, ranging from very small to impressive. Intralesional coagulation results pain, stiffness and distension and calcification of these thrombi, leads to the development of radio-opaque **phleboliths**, which are pathognomonic.

Soft tissue and skeletal **overgrowth** can accompany large VM, **compress the airway** and lead to sleep apnoea. Intraosseus leasion can lead to **pathological fractures**, and intra-articular lesion can lead to **degenerative arthritis**. MRI is the most useful imaging modality.

*Treatment*

Compression therapy, sclerotherapy and surgical resection have all been used with variable success.

## 1.4 Arteriovenous Malformations (AVM)

The pathogenesis is not understood.

*Clinical Findings*

AVM are usually **present at birth**, but are mis-diagnosed due to their innocent appearance. They become more prominent, deeply coloured and a thrill or bruit develops. Bleeding in the surrounding tissues leads to further destruction. CCF can occur in the end stage disease. Limb hypertrophy commonly occurs, and must be treated before 11 years

of age to prevent gross deformity. MRI is used, but CT and angiography can be considered.

*Treatment*
The mainstays of treatment are **combinations** of **embolization, sclerotherapy and resection.** Unlike slow-flow lesions, **complete resection is the goal**, with **frozen section** margin control as rapid recruitment results in recurrence. This is usually very challenging surgery.

*** The remainder of these notes is for completeness and your interest only, and is not examinable ***

## 2. Complex Vascular Malformations

### 2.1 Klippel-Trenaunay Syndrome

This is a **low flow** capillary-lymphatico-venous malformation (**CLVM**) and presents at birth with variable involvement of an extremity (occasionally trunk and pelvis) with soft tissue and skeletal overgrowth. The capillary malformation initially presents as a **macular stain**, but later **haemo-lymphatic vesicles** appear. Enlargement of the **superficial veins** and later the **deep veins** (by **incompetent valves**) occurs. On the lower limb the pathognomonic finding is **the subcutaneous large lateral vein (of Servelle).** Lymphoedema, **macrocysts** and lymphocoeles occur.
Imaging is by **MRI** and importantly, **venography** should be done to assess the venous return from an extremity before undertaking any surgical ablative procedure.

*Treatment*
This is aimed at addressing both the overgrowth and the vascular problems.

**Overgrowth:** Only the lower limb should be treated to **avoid scoliosis**. Custom **orthotics** provide relief from limping initially, and an **epiphysiodesis** is done at around **11 years**. Massive overgrowth is addressed by **staged contour resections** and **selective ablation** (like ray amputation). Wound healing problems are common.

**Vascular problems:** Conservative elastic **compression stockings** are used initially. **Sclerotherapy** (1% SDS), selective **perforator ligation** (document intact deep system before) and **excisional surgery** can be used. Occasionally, radiological **embolization** and excisional surgery with **skin graft** are considered.

### 2.2 Parkes-Weber Syndrome

This is a **high flow** capillary-arterio-venous malformation **(CAVM)** which permeates throughout the limb. It is present at birth. The lower limb is affect more than the upper limb and usually presents as a **pink macular staining limb** with **symmetrical overgrowth.** A **thrill or bruit** is often noted. **CCF** may occur in large lesions. MRI and angiography may demonstrate the lesion.
*Treatment*
In cases of neonatal CCF, **emergency embolization** (which often needs to be repeated) may be life-saving. Overgrowth can be treated by **epiphysiodesis or orthotics** and **repeated selective embolization** may be required for symptomatic control.

# 3. Syndromes with Vascular Malformations as an Integral Part

## 3.1 Bannayan-Riley-Ruvalcaba Syndrome

This is an **overlap syndrome**. It is inherited as an autosomal dominant **(AD)** condition, of PTEN gene on chromosome 10q23. The features include:

- Macrocephaly
- Multiple Lipomas (subcut and visceral)
- **Vascular Anomalies (capillary, venous and AVM – usually minor)**
- Skeletal Abnormalities
- CNS abnormalities (hypotonia, MR, epilepsy)
- Colonic polyps and Propensity for Hashimoto's Thyroiditis

## 3.2 Proteus Syndrome

This syndrome was named after the Greek God, Proteus, who could change form at will (shape shift) to avoid detection. The varied presentations reflect the name given to this syndrome, which lies at the interface of overgrowth syndromes and vascular anomalies. As diagnosis may be difficult, a consensus conference recommended the following **3 diagnostic criteria**:

1. Mosaic or **asymmetrical** distribution
2. **Progressive** course (usually not present at birth)
3. **Sporadic** Occurrence

In addition some of the **"category signs"** must also be present:

- Verrucous linear nevus
- Lipomas
- Macrocephaly (due to calvarial hyperostosis)
- Partial Gigantism of the extemities (overgrowth)
- Cerebriform plantar thickening (aka moccasin feet – a classical sign)

## 3.3 Maffucci Syndrome

This rare syndrome is the constellation of:

1. **Exophytic venous malformations**
2. **Enchondromas**
3. **Bony exostoses**

The presence of bony exostoses and enchondromas is known as Olliers Syndrome.

The condition is **not present at birth** and typically, the osseous lesions begin in early childhood, affecting both axial and extremity bones. The venous malformations develop later, most often on the extremities, but may affect any organ. They sometimes undergo reactive changes, which make differentiation from a vascular tumour difficult.

The most important feature of the syndrome is the **propensity for malignant change**: **chrondrosarcoma** (from an enchondroma – usually low grade) develops in 20-30%, usually in later adulthood. Other tumours include carcinomas of the **ovaries, liver, adrenal and pituitary. Leukemias** have also been described. The exact gene is unknown, but a tumour suppressor gene mutation is suspected.