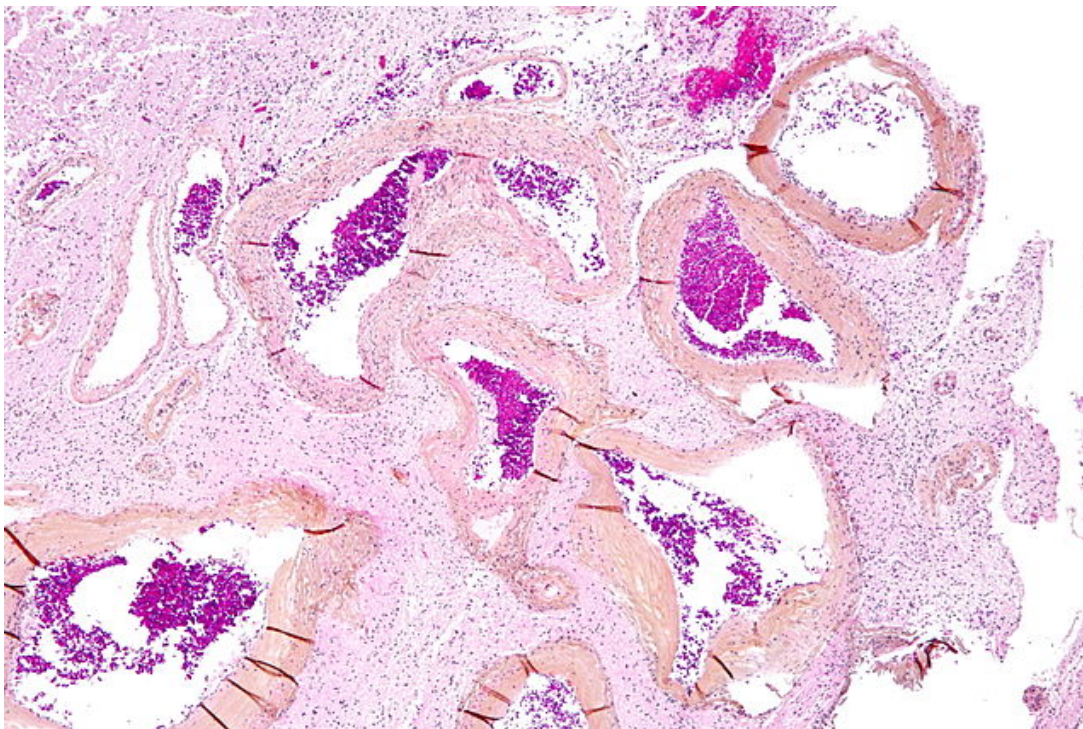


Vascular Malformations: Classification, Investigation and Treatment

[See online here](#)

Vascular malformation has received large attention in recent years due to the frequent occurrence in newborns. Therefore, the clinical features are studied, along with other pathological subtypes, to manipulate the root cause of the disease that seems ordinary but has a great impact on the body.



Introduction

Abnormalities occurring in the fluid carrying vessels in children and adults are referred to as vascular malformations. **Abnormal development of lymph vessels, blood vessels, arteries and veins** leads to disturbances within the normal function of that particular body area through inflammation, pain or bleeding, causing that area to appear as **a congested mass or a cosmetic blemish**.

If the malformation is present at birth, it is regarded as **true vascular malformation**. It develops proportionally as the child grows; it doesn't progress rapidly during childhood nor does it disappear. The malformation may become visible later in life as the flow of blood increases, through abnormal associations between veins and arteries. Poorly developed lymphatic channels or veins can accumulate fluid and **blood**, which may also lead to vascular malformations. They may disturb the normal functions of the **neck and head**, along with other areas of the body, causing cosmetic deformities.

Definition

Vascular malformations — abnormal clusters of blood vessels

Abnormal clusters of blood vessels, known as vascular malformations, form **during fetal development**. They are idiopathic and equally affect **both males and females**. At the time of birth, **lesion is always present**. It might not be visible until days or even years after the child is born, but it usually develops as the child grows. Sometimes, lesions grow quite rapidly. However, it is more typical for them to progress steadily and gradually during infancy. They only diminish when treated.

Examples

Vascular Malformations	
Hemangioma	Non-neoplastic vascular malformations
	Cerebral AVM
	Cavernoma
	Malformation of the aorta: True rings, Double arch, Coarction
	Port wine stains

Classification of Vascular Malformations

Capillary malformations



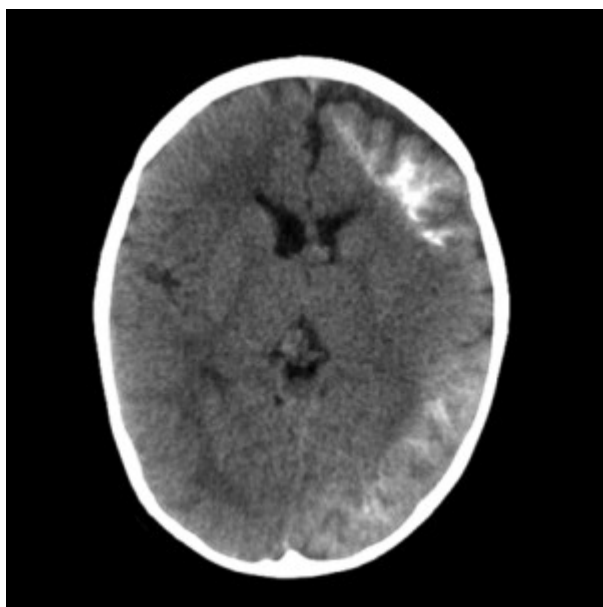
[Image](#): "Port-wine stain" by UnKnown. License: [CC BY-SA 3.0](#)

A **sharply-defined, flat vascular blemish of the skin** is termed capillary malformation and is usually known as a **port-wine stain**. It may appear as speckles forming little islands of color or may cover a huge surface area. These malformations are commonly observed on the **head and neck** but can be present on any part of the body. They are considered as **idiopathic**, but it is assumed that they may arise due to abnormal development of the minute blood vessels of the skin during the embryo phase of life.

The presence of capillary malformations is not linked to medications or drugs taken

throughout pregnancy, nor the experience of environmental abnormalities during that time. Capillary malformations are not caused by a greater number of blood vessels in the affected area. Rather, **the affected blood vessels are quite more dilated than normal**, which causes an **increase in blood flow**. Since the small vessels are very close to the body surface, the increased blood flow causes the skin to appear pink to purple in color.

As the child grows, the affected blood vessels will persist to increase and dilate, which will in turn darken the lesion. Eventually, the clusters of minute, dilated venules give a lumpy impact to the skin. The growth rate of a lesion may vary from person to person and can continue up to the age of 60.



[Image:](#) "CT (without contrast) of the brain of a 20-month-old child with Sturge-Weber syndrome, demonstrating prominent subcortical white matter calcification." by Frank Gaillard. License: [CC BY-SA 3.0](#)

Capillary malformations occurring on the upper eyelid and forehead can be related to lesions of the eye and brain (**Sturge-Weber syndrome**). Capillary malformations occurring in the skin above the spine can be linked with the **Cobb syndrome**, which involves the meninges of the spine. Capillary malformations within this particular area can also be linked with the spine's other abnormalities, which can be diagnosed through an MRI. Capillary malformations situated in the [nose](#), forehead or upper lip can be linked with brain vascular abnormalities and should also be diagnosed.

Lymphatic malformations



[Image](#): "Lymphangioma (Cystic Hygroma)." by Vardhan Kothapalli. License: [Free Art License](#)

These are sponge-like accumulations of clear fluid within abnormal spaces and channels. Collecting surplus fluid from the tissues and emptying it into the venous system by the help of minute vessels is the main job of the lymphatic system. Lymphatic malformations cause a slow transfer of the excess fluid, thus leading to dilation of the vessels and accumulation of the fluid, leading to inflammation of the affected area. It may also cause excessive enlargement of [bones](#) and [soft tissues](#).

These lesions are usually seen in the **axilla and the neck** but can occur in any part of the body. The dilated vessels appear larger than in other body areas. Lesions can be **deep or superficial, diffused or localized**. They gradually increase in size but may sometimes develop rapidly. [Trauma](#) or infection can result in abrupt but transitory enlargement.

Lymphatic malformation is **idiopathic**, but it is assumed that it may occur due to abnormal lymphatic development in the fetus. It is not affected by drugs or environmental exposure during pregnancy.

Venous malformations

Abnormally dilated or developed veins, either deep or superficial, result in venous malformations. These are the **most general form of asymptomatic vascular lesion**. They are naturally present at birth but are rarely seen until weeks or years later, even till adulthood.

Venous malformations **develop gradually and steadily into enlarged veins**. However, infection, surgery, hormonal changes or trauma linked with puberty, menopause or pregnancy may result in swift enlargement. Lesions can be present anywhere in the body, including the skin, the brain, the mucous membrane or the internal organs.

Venous malformations are also considered to be **idiopathic**, but the general opinion is that the deficiency in smooth muscle cells in the veins can be a vital factor. Exposure to drugs and environmental factors during pregnancy have no impact over the venous development of the fetus. Alterations within the genes responsible for the communication between the smooth muscle and endothelial cells have been found.

Arteriovenous malformations (AVM)

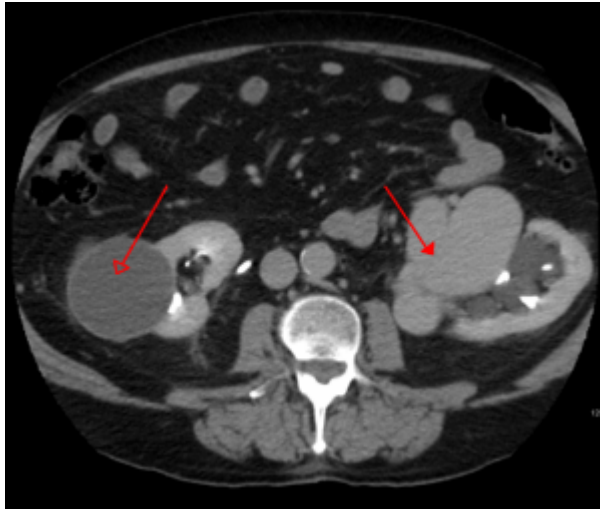


Image: "An arterial venous malformation of the left kidney and a simple cyst of the right kidney" by James Heilman, MD. License: [CC BY-SA 3.0](https://creativecommons.org/licenses/by-sa/3.0/)

The absence of an intervening capillary bed can cause direct associations between the arteries and veins. That leads to localized or diffuse vascular lesions, resulting in an arteriovenous malformation. Localized lesions are likely to be seen on the **neck and head**, often emerge as **vascular light stains** at birth and usually do not expand until early childhood or adolescence.

Lesions are often seen on the **abdomen, chest or limbs**, and they may not be noticeable until later in childhood for they expand with time. Lesions are noticeable in the **inner organs** and may be localized or diffused. They may include organs such as the liver, bowels, brain or the **lungs**. Lesions happening in the brain are a common arteriovenous malformation. They are originally silent, with symptoms development depending upon their rapid enlargement.

The arteriovenous malformations are **idiopathic** in nature and are caused by the irregular formation of the usual arterial-capillary-venous associations during the early developmental stages of life.

Genetic abnormalities that are linked with arteriovenous malformations are **Rasa.1** mutations and **PTEN** mutations. The PTEN gene is involved in typical vascular enlargement. An alteration in PTEN would cause abnormal angiogenesis, which explains the occurrence of AVMs.

Exposure to drugs and environmental factors during pregnancy is not associated with arteriovenous malformations.

Combined vascular malformations

The presence of two or more types of vessel abnormalities in an individual is defined as a combined vascular malformation. Here, the four types of lesion (venous, capillary, arteriovenous or lymphatic) can be combined.

Joint malformations are typically accompanied by overdevelopment of tissues (fat, muscle and skin) **and bones**. While patients may experience some additional skin abnormalities, the usual growth is underneath. The majority of combined malformations occur in the limbs, though other parts of the body can also be involved.

Combined malformations are considered to be idiopathic, but it is assumed that they can

be a result of the abnormal development of various blood vessels in a particular anatomical area during early phases of life.

Clinical Features of Vascular Malformations

Vascular malformations tend to be **clinically dormant until their seemingly sudden activation occurs**. They are generally diagnosed at the time of the first attack or hemorrhage. People with vascular malformation have a **previous history of headaches**. Afterwards, the headaches may catch the form of archetypal migraine. If attacks have occurred one must be careful about seizure history. **Seizures are partial, simple or secondarily generalized**.

Investigations and Diagnosis of Vascular Malformations

Vascular malformations are probably influenced by the **hormonal changes taking place during pregnancy and puberty**. They can also occur as a **result of the accumulation of blood or fluid in abnormally developed lymphatic channels or veins**. Usually, they are present at birth and develop in correlation with the affected child; they neither progress rapidly in childhood, nor do they disappear.

Vascular malformations can become visible later in life as the stream of blood increases within the abnormal associations between veins and arteries. Internal vascular malformations can be diagnosed through an MRI scan.

Treatment of Vascular Malformations

Hemangiomas are different from true malformations because most of the vascular malformations need treatment. Although each vascular malformation cannot be totally eradicated, **recent treatment techniques** help in improvement of the look of the patient and simplify the symptoms of pain, inflammation and bleeding. Numerous adults and children with vascular malformations gain high level of performance and achievement in college, school and different sports.

As true vascular malformations are exceptional and intricate, the best possible outcomes are achieved with diagnosis and treatment at specialized centers. Each patient with a vascular malformation is individual and needs the help of professionals with specific training, experience and skills as well as their way to the highly developed apparatus and resources.

Treatment selection for vascular malformations

The **cerebro-vascular pediatric team** and the **multidisciplinary team** give the most modern and highly developed treatment for all sorts of vascular malformations, which are cautiously examined for the right problem, location, age and symptoms of the patient. These treatment methods are:

Embolization

Embolization is an invasive method where “glues” or particles are used to **close the abnormally developed blood vessels**. Even though this method can seal the abnormality in few patients, there are cases where the malformation needs to be

eradicated surgically. Radio surgery is also an option, with the use of certain types of radiation. In such cases, embolization prior to each of these techniques can minimize the malformation and assist in decreasing the risk of blood loss if surgical intervention is necessary.

Laser treatment

It is used mostly for **treating venous malformations that are superficial or where a deep lesion contains a superficial component**. Physicians must use their extensive knowledge in navigating the type of therapy that is best for each patient. The lasers used for this type of treatment efficiently decrease the bluish stains of the inside layer of the skin, the lips and the mouth.

A cooling device is used in tandem with the laser, so that the skin is feasibly safe and intact for the curing of the vascular malformation underneath. Recently, the laser treatment approach is often performed alongside sclerotherapy as well as surgery for a useful treatment of composite venous malformations.

Sclerotherapy

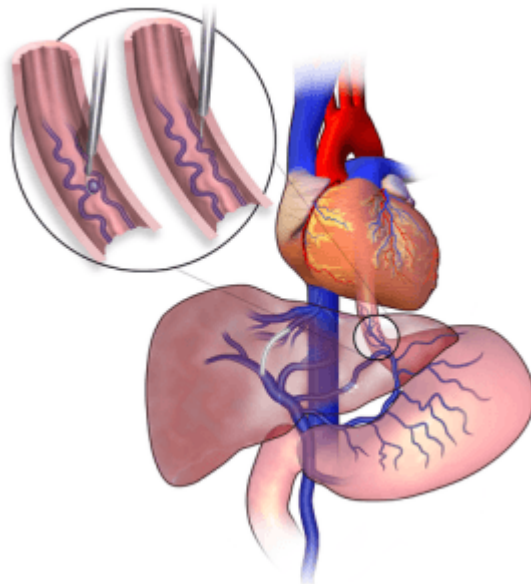


Image: "Sclerotherapy" by BruceBlaus. License: [CC BY-SA 4.0](https://creativecommons.org/licenses/by-sa/4.0/)

This therapy is frequently employed in **treating vascular malformations as well as lymphatic malformations**. A solution is injected into the anomalous vessel via the skin. The solution can be among one of few that are competent to block the vessel so the stream of blood is discontinued. This leads to deterioration of the vessel and rerouting the blood flow toward healthier vessels.

Surgery

Surgical elimination of a vascular malformation is performed in stages, although it may work as a single operation as well. Recently, physicians have perfected a treatment in which the lesion is initially cured with sclerotherapy and later, within 48 hours, the malformation is surgically eliminated.

Using sclerotherapy reduces the risk of rigorous blood loss during surgery and lowers the number of required surgical procedures. The benefits of this joint treatment are

tremendous, especially in eradicating injuries that were formerly considered untreatable.

Additionally, experience shows that large malformations cured with sclerotherapy evade bulky disfiguration marks a lot better than ordinary surgery performed by itself. In addition, microcystic (small cysts) lesions, often reappear after surgery, and so several procedures are typically necessary. With sclerotherapy and a method known as **gravity technique**, this treatment has been completely transformed.

For vascular malformations, surgery can be a choice only after embolization.

With this combination, surgery is performed 48 hours after embolization. However, if the condition is widespread, it is significant to operate only on enough tissue to prevent reappearance, but evade disfigurement.

High-yield Notes of Vascular Malformation

	Vascular Tumors (Hemangiomas)	Vascular Malformations
Presence at birth	Not present at birth but appear postnatally	There are errors of morphogenesis and are presumed to occur during intrauterine life → Most present at birth
Natural history	<p>Phases:</p> <ol style="list-style-type: none"> Proliferating: Grow rapidly during the first year Involuting: Slow during childhood over 2-6 years. Involuted: By the age of 10 years 	Proportionate growth; can expand
Cellular	Endothelial hyperplasia	Normal endothelial turnover
Skeletal changes	<ul style="list-style-type: none"> Occasional mass effect on adjacent bone Rare hypertrophy 	<ul style="list-style-type: none"> Slow-flow: Distortion, hypertrophy or hyperplasia Fast-flow: Destruction, distortion or hypertrophy

High-yield Notes of Vascular Tumors

Hemangioma of Infancy (Strawberry Hemangioma)

Most common tumor of infancy with F : M ratio (3:1)

Clinical features

Description:

- Soft bright red (more violaceous when deeper), sharply demarcated, raised lesions
- Appearing in first 2 months → rapidly expanding → then involuting by age 5-9 years

Life cycle in 3 phases:

Proliferation stage	<ul style="list-style-type: none"> Initial notion as an erythematous, macular patch
Involution stage	<ul style="list-style-type: none"> Changes/ deepens its color and grows faster than the growth of the child <ul style="list-style-type: none"> Mostly started by 12 months of age Slow process, completes by the age of 5 to 9 years
Plateau phase	<ul style="list-style-type: none"> No growth lasts from 9 to 12 months of age

Distribution:

- Lesions are usually **solitary and localized or extend over an entire region**
- **Head & neck 50 % /trunk 25 %**. Face, trunk, legs, oral mucous membrane

Special presentations: deep hemangioma (formerly cavernous hemangioma)

- Consist of **dilated vascular spaces** with **thin-walled endothelial cells**
- Presentation as **soft blue, compressible mass up to a few centimeters in size**
- Cavernous hemangiomas may appear on the **skin, viscera, deep tissues, mucosa** → If it involves the larynx it can cause obstruction
- **Less likely to regress spontaneously than capillary hemangiomas**
- Cavernous hemangiomas of **brain & viscera** are associated with **von Hippel-Lindau disease**

Treatment

- **No intervention** is the best: because spontaneous resolution gives the best cosmetic results
- Treatment is indicated if a hemangioma 1) Ulcerates 2) Obstructs vital structures 3) may become life-threatening in the near future
 - **Surgical:** 1) Continuous wave or pulsed dye laser 2) Cryosurgery
 - **Medical:** 1) Intralesional and systemic high-dose glucocorticoids 2) Interferon α (IFN- α) 3) Propranolol

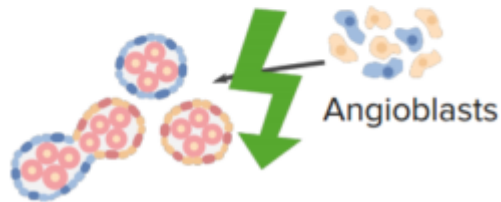
Hemangioma		
Low Flow	High Flow	Rare
Capillary (CM)	COMBINED arteriovenous (AVM)	Capillary-lymphatic (CLM)
Lymphatic (LM)		Capillary venous (CVM)
Venous (VM)		Lymphatico-venous (LVM)
Arterial (AM)		Capillary-lymphatico-venous

Pyogenic Granuloma

- Very common **solitary eroded vascular nodule** that bleeds **spontaneously or after minor trauma**
- Appears as **bright red, dusky red, violaceous, or brown-black papule with a collar of hyperplastic epidermis at the base**

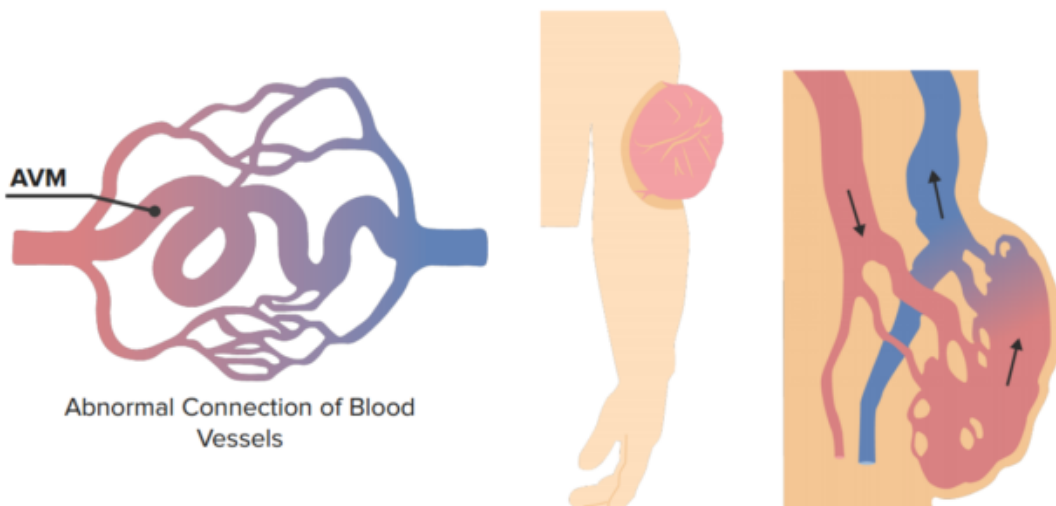
Arteriovenous Malformations (AVM)

Non-neoplastic vascular malformations



Blood islands

- Present at birth, do not go through a rapid proliferative phase and do not involute
- Grow with the patient
- Approximately 31 % are found in the head and neck
- Are thought to result when there is interruption at a particular stage of development of a vessel
- Type of malformation depends on the stage which is normal morphogenesis is interrupted
- Abnormal connection between arteries and veins
- Bypassing the capillary system
- Can appear in any location
- Mostly asymptomatic
- Can cause intense pain, bleeding or lead to other serious medical problems
- Dilatation with time
- Subtype cerebral AVM

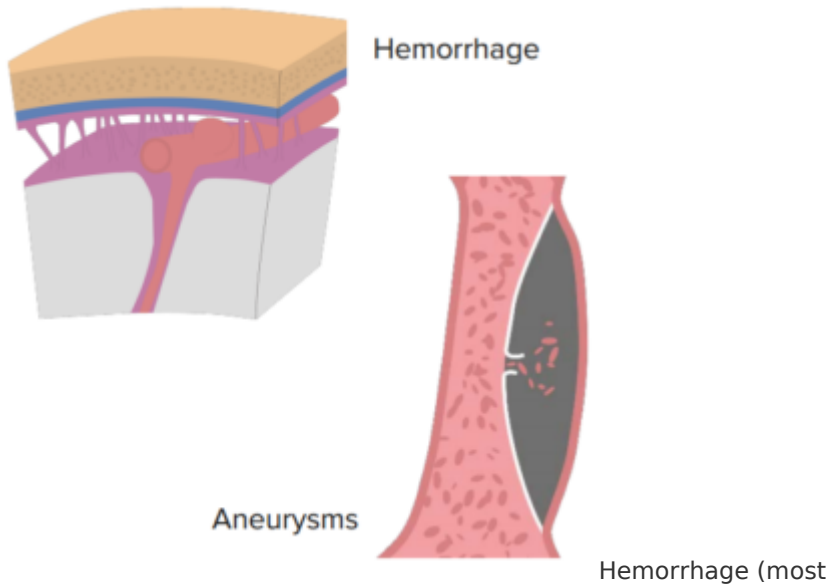


Cerebral AVM

- Classification via Spetzler-Martin Grading System
- The higher the AVM score, the more difficult is treatment

Spetzler-Martin Grading System for AVMs		
Size of AVM	Small (< 3 cm)	1
	Medium (3–6 cm)	2
	Large (> 6 cm)	3
Eloquence of adjacent brain	Non-eloquent	0
	Eloquent	1
Pattern of venous drainage	Superficial only	0
	Deep component	1

Clinical representation



- common subarachnoid and inter-cerebral)
- Seizures
- Mass effect (increased intracranial pressure)
- Progressive neurologic deficit
- 7 % of patients with AVMs present with associated aneurysms

Diagnosis

- CT-Scan
- Ct angiography
- MRI scan
- MRI angiography
- Cerebral angiography

Cavernomas

Types of cavernomas

1. Cavernomas angioma
2. Cavernomas hemangioma

Symptoms

- Seizures
- Progressive neurological deficit
- Hemorrhage (usually with the brain itself)

- > 50 % asymptomatic

Diagnosis

- CT
- MRI — hemosiderin ring (*"salt and pepper" appearance*)
- Angiography

Vascular Malformations: Capillary Malformation (CM)

Port-Wine Stain (Nevus flammeus)

Clinical features

- **Irregularly shaped, red or violaceous, macular CM** that is **present at birth** and **never disappears spontaneously**
- Tends to increase in size with the growth of the child → becomes raised & nodular, causing significant disfigurement.
- **Distribution:**
 - Most commonly **involve the face** in the **distribution of the trigeminal nerve**, usually the superior & middle branches
 - **Mucosal** involvement of **conjunctiva & mouth** may occur

Syndromic CM

- **Sturge-Weber syndrome (SWS)** is the association of **Port-Wine Stain** in the trigeminal distribution with:
 - **Eye:** Vascular malformations in the eye and
 - **Brain:** Leptomeninges & superficial calcifications of the brain
- **Klippel-Trénaunay-Weber syndrome:** May have an associated **PWS overlying the deeper vascular malformation of soft tissue & bone**

Spider Angioma (Spider Nevus)

Consists of a **bright red central papule (dilated central arteriole)** **surrounded by a telangiectatic network of dilated capillaries**. Associated with **hyperestrogenic states**, such as pregnancy, in patients receiving estrogen therapy, e.g., oral contraceptives, or in those with the hepatocellular disease. May regress spontaneously.

Cherry Angioma (Cherry Hemangiomas) Senile Hemangiomas

Appears during the third or fourth decade of life. **Does not regress spontaneously**; their number often increases with age. Always **cutaneous and not found on the mucosa or deep tissues**.

Description: Sharply circumscribed areas of congested capillaries & post-capillary venules in the papillary dermis

Review Questions

The correct answers can be found below the references.

1. A 3-month-old infant has soft, bright-red, sharply-demarcated, raised lesions over the head and neck. Hemangioma of infancy was diagnosed. The best next step in the management of the infant is... ?

- A. No intervention, as it will involute by the age of 5 years
- B. Laser treatment
- C. Sclerotherapy
- D. Surgical removal

2. Sturge-Weber syndrome (SWS) is the association of Port-Wine Stain in the trigeminal distribution with... ?

- A. Intraventricular hemorrhage
- B. Cavernous hemangiomas of the brain
- C. Vascular malformations in the eye
- D. Vascular malformation of soft tissue & bone

3. Vascular tumors (hemangiomas) are characterized by their presence...

- A. ...in post-natal infants.
- B. ...in intrauterine life.
- C. ...at birth.
- D. ...in adulthood.

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High-yield notes of Vascular Malformation & Tumor for USMLE was kindly provided by www.mediconotes.com

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Correct answers: 1A, 2C, 3A

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Notes