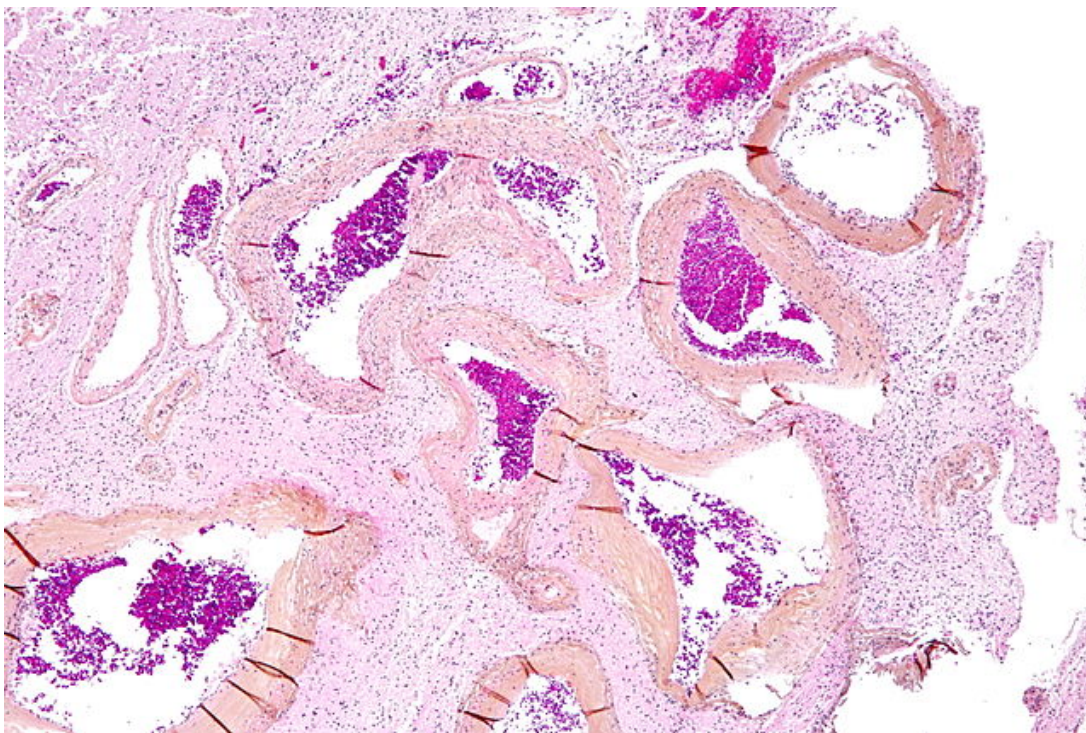


Vascular Malformations: Classification, Investigation and Treatment

[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. The **abnormal development of lymph vessels, blood vessels, arteries, and veins** leads to disturbances in the normal function of these structures through inflammation, pain, or bleeding, causing such areas to appear as **a congested mass or cosmetic blemish**.

Malformations present at birth are considered **true vascular malformations**. A true vascular malformation develops proportionally as the child grows; it does not progress rapidly during childhood or 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 lead to vascular malformations. They may impair 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 **males and females**. At birth, the **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. Occasionally, lesions grow 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 arteriovenous malformations (AVM)
	Cavernoma
	Malformation of the aorta: True rings, Double arch, Coarctation
	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 a 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 could arise due to the abnormal development of the minute blood vessels of the skin during the embryonic phase of life.

The presence of capillary malformations is not linked to drugs taken or exposure to environmental factors during pregnancy. Capillary malformations are not caused by a

greater number of blood vessels in the affected area. Rather, **the affected blood vessels are 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 persist and increase and dilate, which in turn darkens the lesion. Eventually, the clusters of minute, dilated venules give a lumpy feel to the skin. The growth rate of a lesion may vary from person to person and can continue up to the age of 60 years.

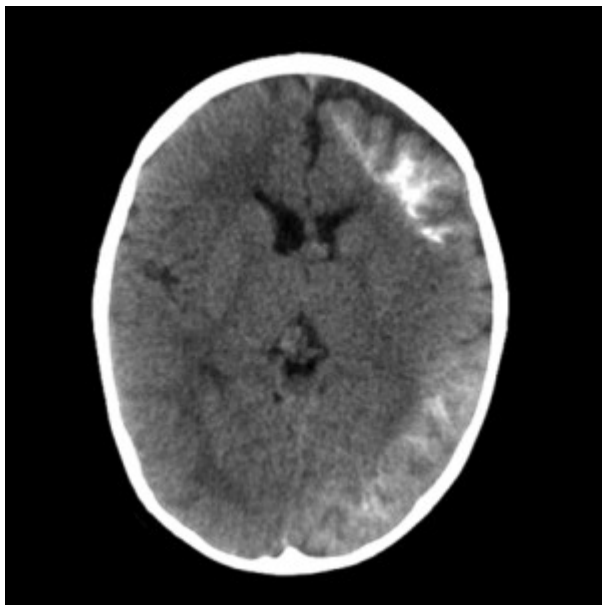


Image: CT (without contrast) of the brain of a 20-month-old child with Sturge-Weber syndrome, demonstrating prominent subcortical white matter calcification. By: Frank Gaillard. License: [CC BY-SA 3.0](https://creativecommons.org/licenses/by-sa/3.0/)

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 to the **Cobb syndrome**, which involves the meninges of the spine. Capillary malformations within this particular area can also be linked with other spinal abnormalities, which can be diagnosed with an MRI. Capillary malformations situated in the nose, forehead, or upper lip are linked to brain vascular abnormalities and should also be diagnosed.

Lymphatic malformations



[Image](#): Lymphangioma (Cystic Hygroma). By: Vardhan Kothapalli.
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These are sponge-like accumulations of clear fluid within abnormal spaces and channels. The main function of the lymphatic system is to collect surplus fluid from the tissues and empty it into the venous system via minute vessels. Lymphatic malformations cause a slow transfer of the excess fluid, leading to dilation of the vessels and accumulation of the fluid. This results in the inflammation of the affected area. Lymphatic malformations may also cause excessive enlargement of the [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 and/or diffused or localized**. They gradually increase in size but may sometimes develop rapidly. [Trauma](#) or infection can result in abrupt but transitory enlargement.

A lymphatic malformation is considered **idiopathic** but is assumed to 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. Venous malformations are the **most general form of asymptomatic vascular lesions**. They are naturally present at birth but are rarely seen until weeks or years later, even until adulthood.

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

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

Arteriovenous malformations (AVM)



Image: An arteriovenous 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. This leads to localized or diffuse vascular lesions, resulting in an AVM. Localized lesions are likely to be seen on the **neck and head**, often emerging as **vascular light stains** at birth, and they usually do not expand until early childhood or adolescence.

Lesions are often seen on the **abdomen, chest, or limbs**, and may not be noticeable until later in childhood as 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 [lungs](#). Lesions occurring in the brain are common AVMs. They are originally silent, with symptom development depending on their rapid enlargement.

AVMs are **idiopathic** 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 AVMs are the **Rasa.1** 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 AVMs.

Combined vascular malformations

The presence of 2 or more types of vessel abnormalities in an individual is defined as a combined vascular malformation. Here, the 4 types of lesions (venous, capillary, arteriovenous, or lymphatic) can simultaneously occur.

Combined malformations are typically accompanied by the 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, although other parts of the body can also be involved.

Combined malformations are considered idiopathic but could be a result of the abnormal development of various blood vessels in a particular anatomical area during the 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. Patients with vascular malformations have a **previous history of headaches**. Afterward, the headaches may resemble an archetypal migraine. If attacks have occurred, a detailed seizure history is crucial. **Seizures are partial, simple, or secondarily generalized**.

Investigations and Diagnosis of Vascular Malformations

Vascular malformations are probably influenced by the **hormonal changes that take 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 relation to the affected child; they neither progress rapidly in childhood nor disappear.

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

Treatment of vascular malformations

Hemangiomas differ from true malformations because most of the vascular malformations need treatment. Although the vascular malformations cannot be totally eradicated, **recent treatment techniques** help in improving the patient's appearance and alleviating the symptoms of pain, inflammation, and bleeding. Several adults and children with vascular malformations achieve a high level of performance in college, school, and different sports after treatment.

As true vascular malformations are exceptional and intricate, the best possible outcomes are achieved with diagnosis and treatment at specialized centers. Patients with vascular malformations need individualized treatment from professionals with specific training, experience, and skills, in addition to highly developed equipment and care.

Treatment selection for vascular malformations

The **cerebrovascular pediatric team** and the **multidisciplinary team** provide the most modern and highly developed treatment for various types of vascular malformations, evaluating for the right signs and symptoms, location, and demography of the patient. These treatment methods include:

Embolization

Embolization is an invasive method where 'glues' or particles are used to **close the abnormal blood vessels**. Although this method can seal the abnormality in a few patients, there are cases where the malformation needs to be surgically removed. Radiological 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

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

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

Sclerotherapy

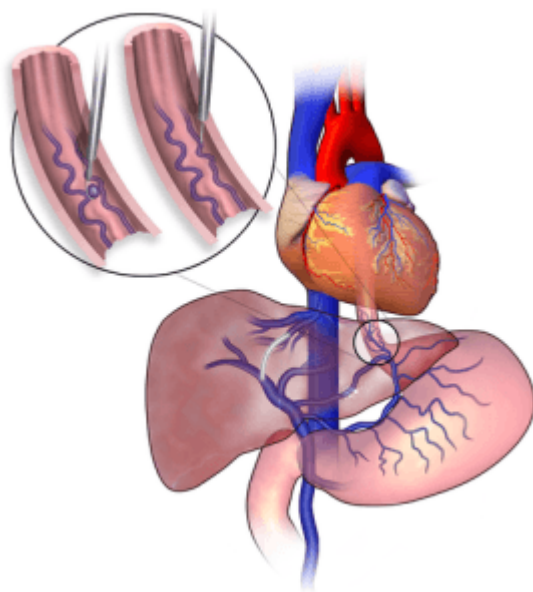


Image: Sclerotherapy. By: BruceBlaus. License: [CC BY-SA 4.0](https://creativecommons.org/licenses/by-sa/4.0/)

This technique is frequently employed in **treating vascular and lymphatic malformations**. A solution is injected into the anomalous vessel via the skin. The solution is able to block the vessel so that blood flow is discontinued. This leads to the deterioration of the vessel and the rerouting of blood toward healthier vessels.

Surgery

Surgical elimination of a vascular malformation is performed in stages, although it may also be performed as a single operation. Recently, physicians have developed 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 profound blood loss during surgery and lowers the number of required surgical procedures. The benefits of this combined treatment are notable, especially with regard to the eradication of injuries that were formerly considered untreatable.

Additionally, experience shows that large malformations cured with sclerotherapy prevent bulky disfiguration marks compared to surgery alone. In addition, microcystic (small cysts) lesions often reappear after surgery, and so several procedures are typically necessary. With sclerotherapy and a method known as the **gravity technique**, this treatment has been 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 important to operate only on enough tissue to prevent recurrence and disfigurement.

Important Notes about Vascular Malformation

	Vascular Tumors (Hemangiomas)	Vascular Malformations
Presence at birth	Not present at birth but appear postnatally	These are errors of morphogenesis and are presumed to occur during intrauterine life → Mostly present at birth
Natural history	Phases: 1. Proliferating: Grows rapidly during the first year 2. Involuting: Slow during childhood over 2-6 years 3. 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

Important Notes about Vascular Tumors

Hemangioma of infancy (Strawberry hemangioma)

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

Clinical features

Description:

- Soft bright-red (more violaceous when deeper), sharply demarcated, raised lesions
- Appearing in the 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 appearance 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 starts by 12 months of age • Slow process, is completed by the age of 5-9 years
Plateau phase	<ul style="list-style-type: none"> No growth, lasts from 9-12 months of age

Distribution:

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

Special presentations: deep hemangioma (formerly cavernous hemangioma)

- Consists of **dilated vascular spaces** with **thin-walled endothelial cells**
- Presents as a **soft, blue, compressible mass up to a few centimeters in size**
- Cavernous hemangiomas may appear on the **skin, viscera, deep tissues, and mucosa** → If the larynx is involved, an obstruction may result
- **Less likely to regress spontaneously than capillary hemangiomas**
- Cavernous hemangiomas of the **brain** and **viscera** are associated with **von Hippel-Lindau disease**

Treatment

- **No intervention** is the best: because spontaneous resolution yields the best cosmetic results
- Treatment is indicated if a hemangioma 1) ulcerates 2) obstructs vital structures or 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

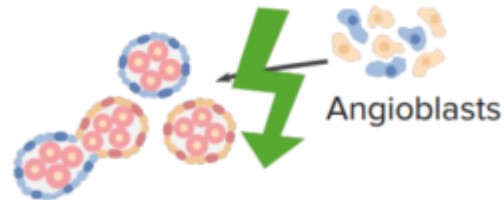
Hemangioma		
Low-flow malformations	High-flow malformations	Rare malformations
Capillary	Combined arteriovenous	Capillary-lymphatic
Lymphatic		Capillary-venous
Venous		Lymphatico-venous
Arterial		Capillary-lymphatico-venous

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

Non-neoplastic vascular malformations



Blood islands

Image: Angioblasts form inappropriate 'islands' of cells that blood flows through in AVMs. By: Lecturio

- Present at birth, do not go through a rapid proliferative phase and do not involute
- Grow with the patient
- Approx. 31% are found in the head and neck
- Are thought to result when there is an interruption at a particular stage of vessel development
- The type of malformation depends on the stage at which 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

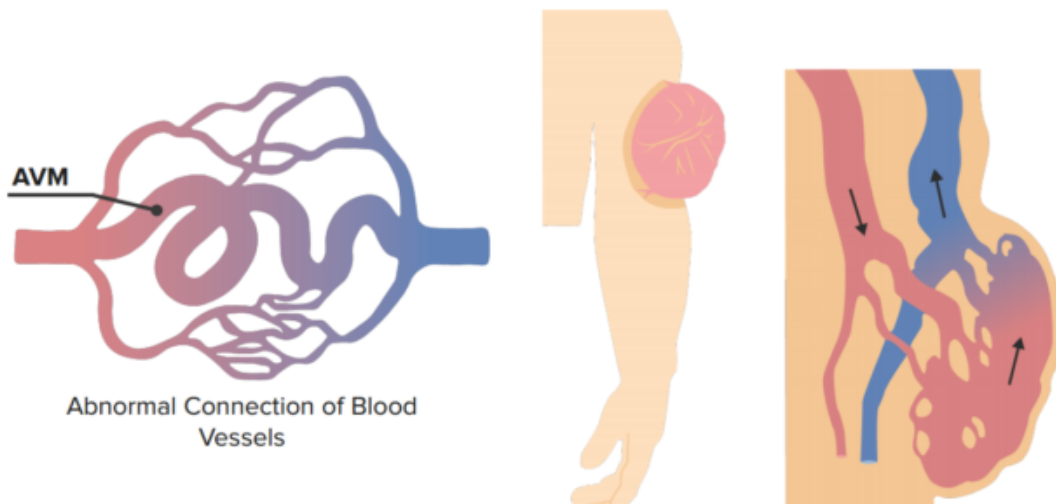


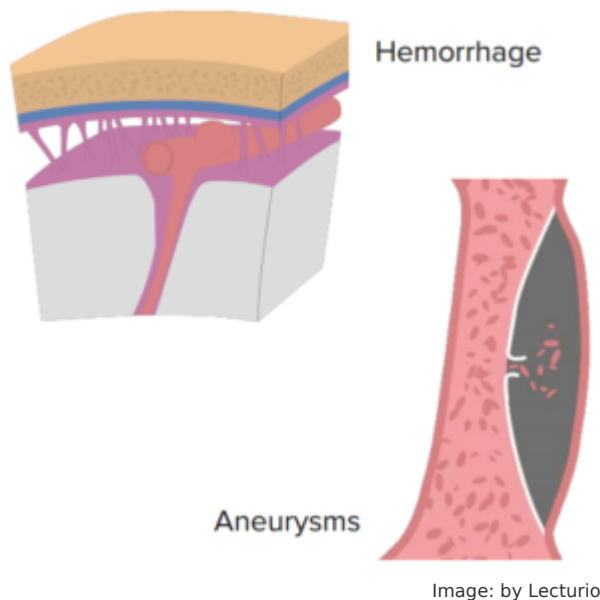
Image: Images show an inappropriate connection between the arterial and venous vessels resulting in enlargement with a blood-filled skin lesion. By: Lecturio

Cerebral AVM

- Classification via the Spetzler-Martin grading system
- The higher the AVM score, the more difficult the 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



- Hemorrhage (subarachnoid and intercerebral are the most common)
- Seizures
- Mass effect (increased intracranial pressure)
- Progressive neurologic deficit
- 7% of patients with AVMs present with associated aneurysms

Diagnosis

- CT scan
- CT angiography
- MRI
- MR angiography
- Cerebral angiography

Cavernomas

Types of cavernoma

1. Cavernous angioma

2. Cavernous hemangioma

Symptoms

- Seizures
- Progressive neurological deficit
- Hemorrhage (usually within the brain)
- > 50% asymptomatic

Diagnosis

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

Vascular Malformations: Capillary Malformation

Port-wine stain (Nevus flammeus)

Clinical features

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

Syndromic capillary malformation

- **Sturge-Weber syndrome** is the association of **port-wine stain** in the trigeminal distribution with:
 - **Eye:** Vascular malformations in the eye
 - **Brain:** Leptomeninges and superficial calcifications in the brain
- **Klippel-Trénaunay-Weber syndrome:** May have an associated **port-wine stain overlying the deeper vascular malformation of soft tissue and 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 hepatocellular disease. May regress spontaneously.

Cherry angiomas (Cherry hemangiomas) and Senile hemangiomas

Appear during the 3rd or 4th decade of life. **Do not regress spontaneously**; numbers often increase with age. They are always **cutaneous and are not found on the mucosa or deep tissues**.

Description: Sharply circumscribed areas of congested capillaries and post-capillary

venules in the papillary dermis.

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

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Notes