VASCULAR BIRTHMARKS: PART 2

Vascular Malformations

Samuel Negash MD
Pediatric Surgery Resident
Addis Ababa University

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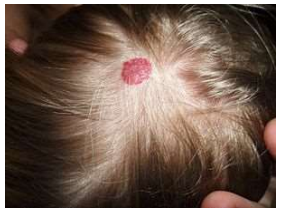
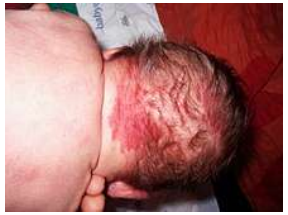
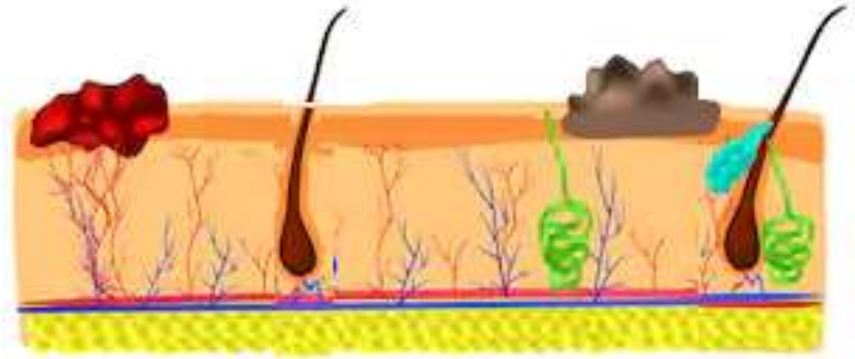
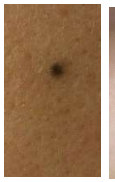
TIKUR ANBESSA

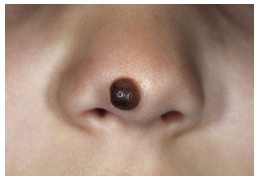
What are birthmarks?

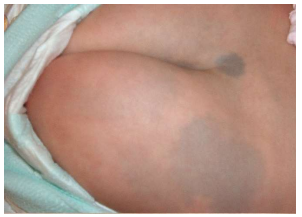












Benign irregularity on the **skin** present at **birth** or shortly afterwards

Introduction

- Occur in **1.5% of the population** (M:F = 1:1)
- **Notorious reputation for wide variety**: location, extent, independent or mixed lesion, unpredictable clinical course
- Remains the **most difficult and confusing** diagnostic and therapeutic clinical entity among vascular disorders
- **Erratic response** to treatment with high recurrence

OUTLINE

1. Embryology
2. Classification
3. Approach
4. Low flow lesions
5. High flow lesions

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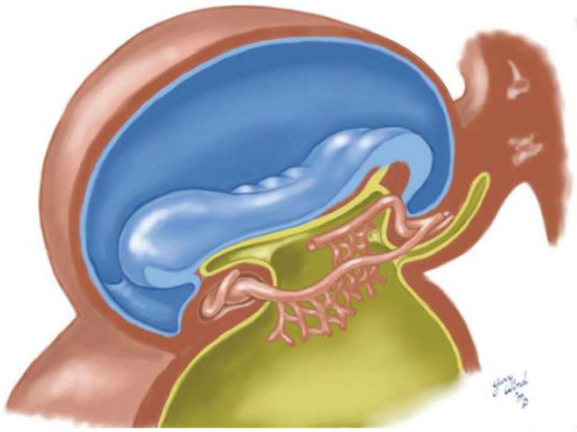
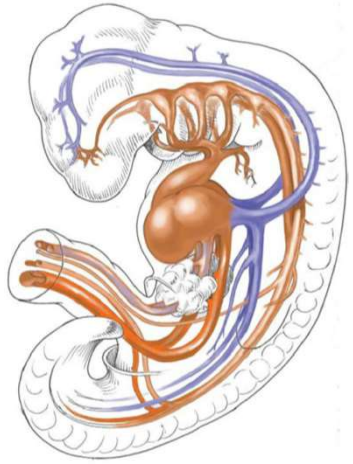
Development of vascular system



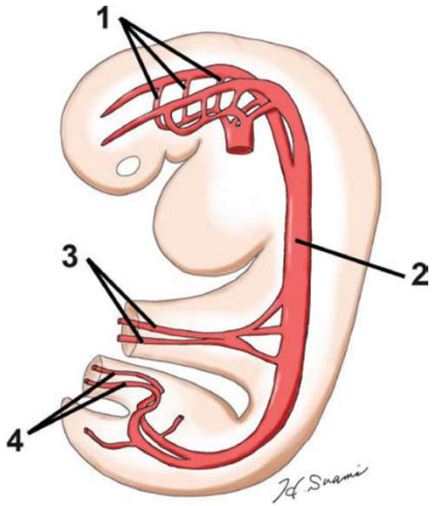


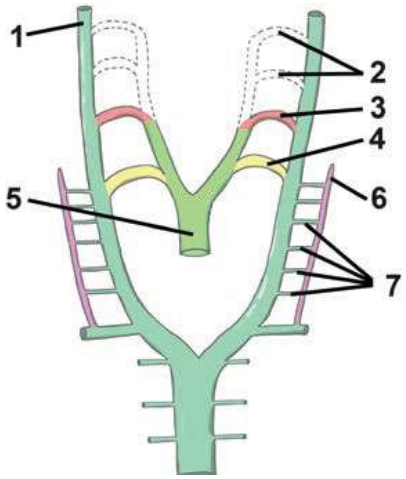
- **3rd week**, The first two sets of primordial vessels attach to the ends of the newly fused heart tube**. 4th week,** aortic fusion has begun. arches are forming, the umbilical vessels and the cardinal veins are formed,

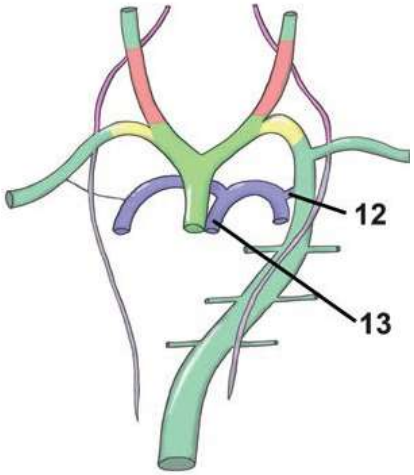
Development of vascular system

- **Vasculogenesis** - vascular channels from mesoderm (angioblast)
- **Angiogenesis** – new vessels from preexisting vessels, Remodeling into arteries and veins take place
  - Regulators – heart beat and blood flow
  - VEGF, basic FGF-2, and angiopoietin 1 (Ang-1)…
- **lymphatics** - mid-gestation after the blood vasculature forms.
  - Form from venous endothelium or independent progenitors

Arterial system Development

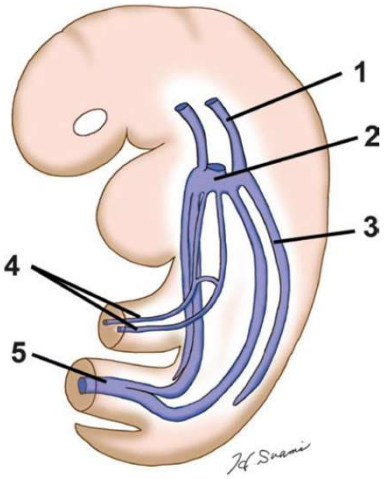


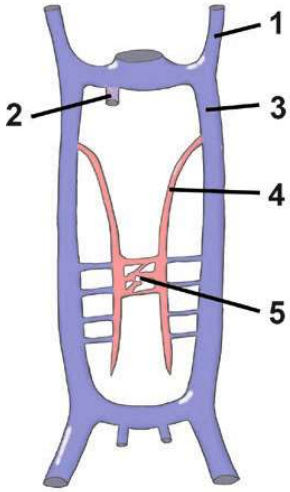


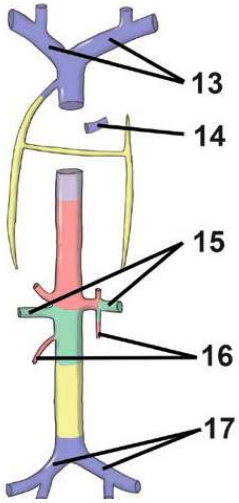


4th wk = aortic sac, aortic arches, dorsal aorta, vitelline arteries, and umbilical arteries. Further development 6th – 8th wk.

Venous system Development

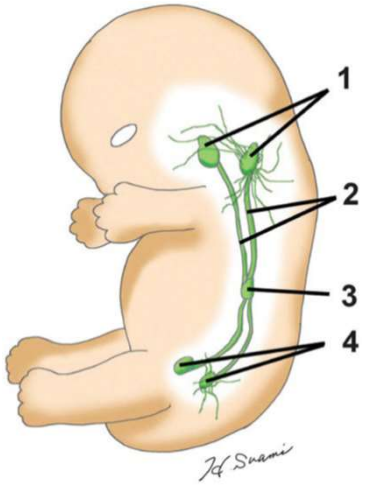


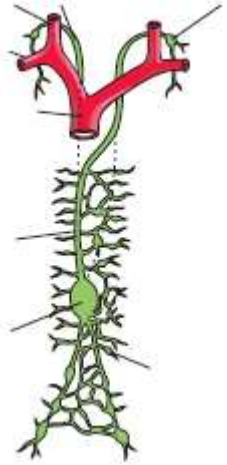


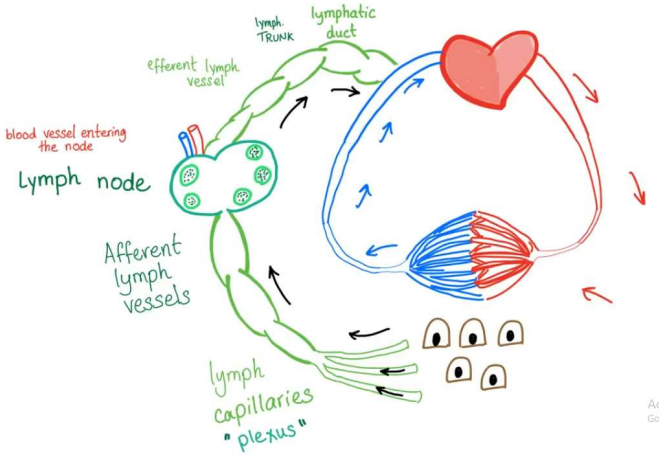


4th wk = anterior cardinal vein, sinus venosus, posterior cardinal vein, vitelline veins, and umbilical vein. Further development 5th -7th wk.

Lymphatic system Development







7th wk = 6 Lymph sacs (jugular, retroperitoneal, cysterna chili, iliac)
Later, lymphatic vessels connect to sacs and pass along main veins.

Pathogenesis

- **Defects in** any of the proteins involved in the **regulation**

↓

Abnormal channels

↓

Subsequently expand

- *Anomalies of origin, course, number, length, stenosis/obstruction (aplasia, hypoplasia) dilatation (ectasia / aneurysm), valves, communication (AVF), persistence of embryonal vessel*

Pathophysiology

- **Extratruncular stage**: undifferentiated vasculature (angioblast)
  - **Growth** - Rapid, stimulation (time, hormone, trauma, infection, thormbus)
  - **Risk of recurrence** – invasion of surrounding structures
  - **Mechanical impact** - Compression and infiltration
- **Truncular stage**: differentiated to artery, lymph, vein
  - No potential to proliferate, either immature or hyperplastic
  - **More Hemodynamic impact** (highest in AVF)
  - **Secondary organ impact** (stimulating bone growth)

Etiology

- **Congenital**
  - Usually sporadic
  - **Teratogens** and **maternal disease** have been associated
  - **Genetic mutation:** dysfunction in endothelial regulation
- **? Acquired AVM**
  - DVT, ischemia and trauma >> upregulation of growth factors

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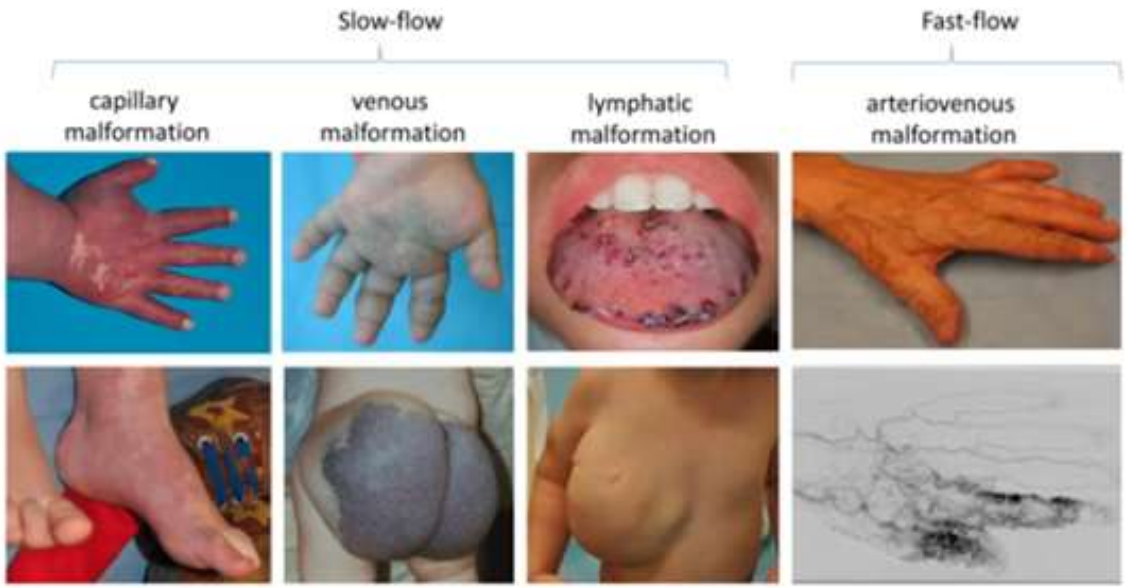
Common misconceptions

- All **vascular birthmark's** are not " hemangiomas"
- All **vascular malformations** are not "AVMs"

Classification systems

- **Old classification**: Virchow, others
  - didn't consider etiology, anatomy, and pathophysiology
- **Hamburg** (1988): limited to vascular malformations
  - removed eponyms
- **ISSVA** (1996, 2014, 2018): all vascular anomalies
  - classification based on flow characteristics

Simple Malformations



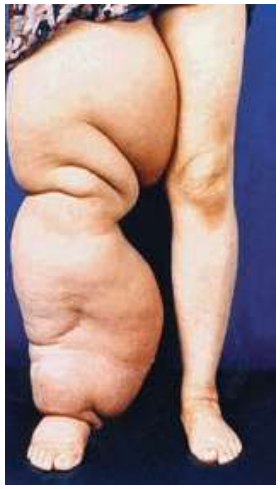
ISSVA: **Simple**, Combined, of major named vessels, Syndromic

Combined Malformations

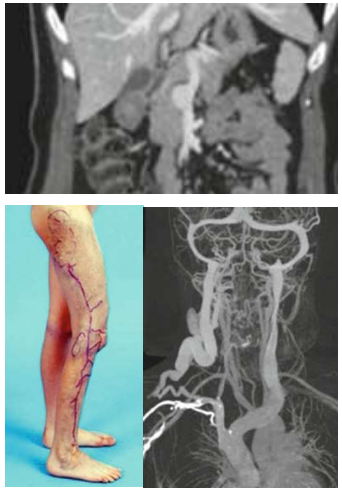
ISSVA: Simple, **Combined**, of major named vessels, Syndromic

Malformations of major named vessels

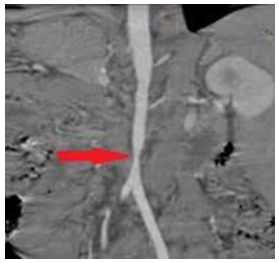
Lymphatic

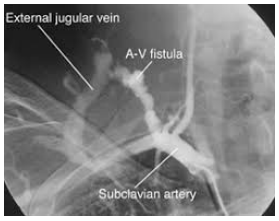


Venous



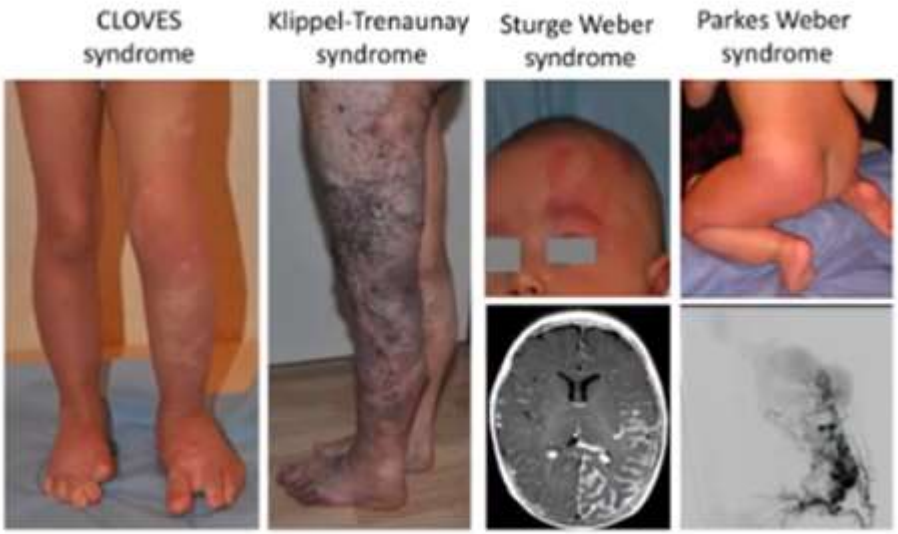
Arterial





ISSVA: Simple, Combined, **of major named vessels**, Syndromic

Syndromic Malformations



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Presentation

- **Congenital**, but may not become apparent until adulthood
- Similar appearance weather on **skin, mucosa or viscera**
  - Flat/raised
  - discoloration (pink/red/purple/blue)
- **Pain**, **limb over growth**
- **Can confuse with infantile hemangiomas** especially when they have a significant fast-flow component

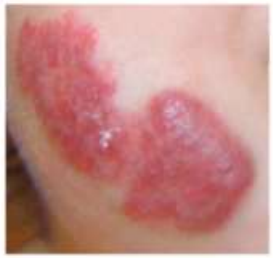
First step is differentiating

	Infantile Hemangioma	Vascular Malformation
Pathology	Tumor	Malformation
Epidemiology	3x more in girls	Equal
Age	Appear in neonatal; infant/child	Present at birth, may appear in adult
Growth	Rapidly Proliferate in infancy Involute over 5-10 years	Grow with child (proportional) Grow will stimulation
Clinical exam	red, raised lesion bluish if deeper Occasional ulceration & bleeding	CM = flat ; LM = transilluminate VM = compressible, Valsalva/dependent AVM = warm, thrill, bruit

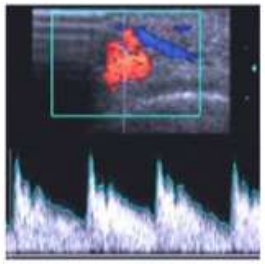
Clinical examination

- **General** - well-lit environment, standing and supine positions
- **Inspection and palpation** of all skin surfaces, including the genitalia, should be performed in a environment.
- **Auscultation** (for bruit) and palpation (for thrills)
- The **limb elevation** test should be performed in extremities
- **hemodynamic impact** on the distal and proximal vascular systems, and on central cardiovascular system

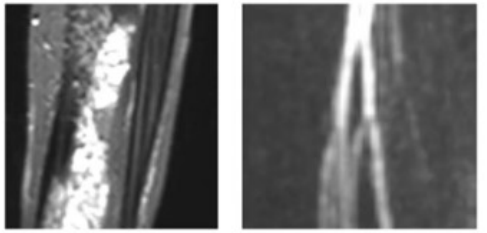
Diagnostic approach



Clinical examination

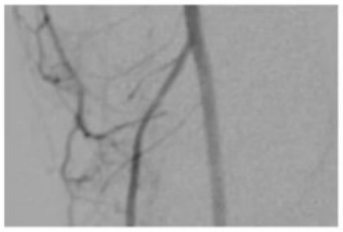


Sonography

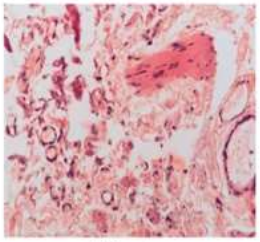


Magnetic resonance
with and without angiography

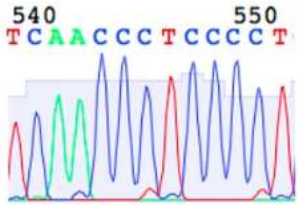
Blood tests



Catheter angiography



Biopsy

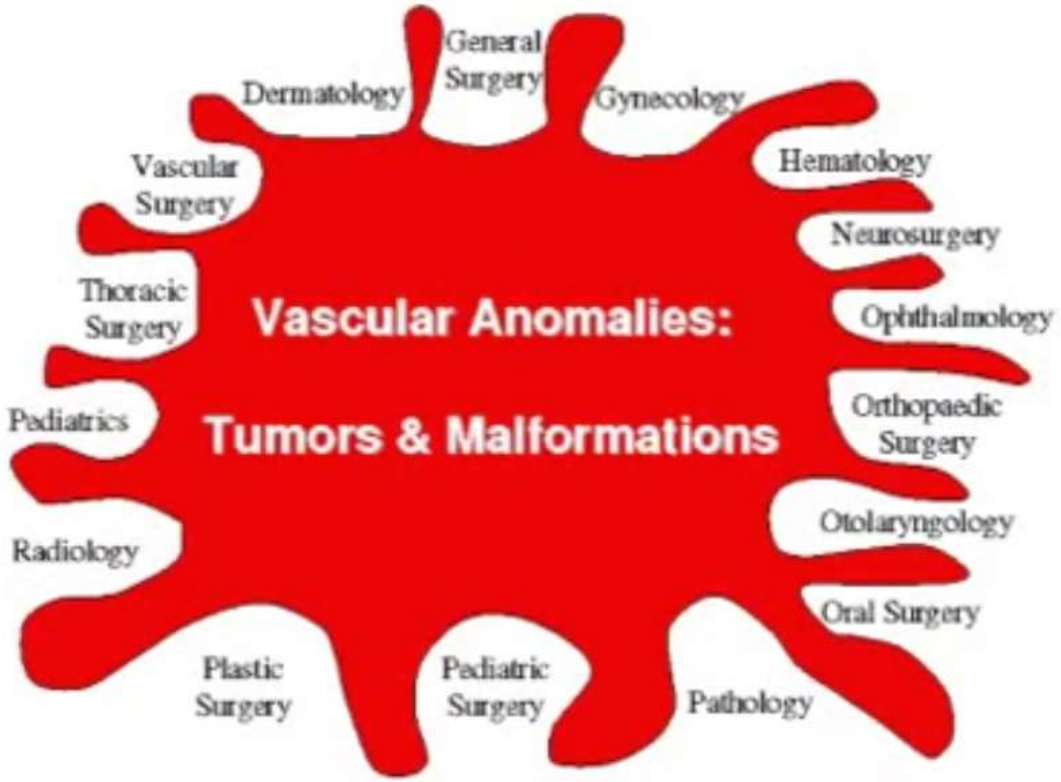


Genetic testing

Imaging

- **Non invasive**: rule out hemangioma, identify type, other malformation
  - **Venous arterial duplex**– Differentiate fast/slow flow. fluid filled cyst.
  - **MRI (T2 weighted)–** MRI allows assessment of lesion extent, severity, Relationship with the surrounding tissues and organs
- **Invasive**: to plan surgery (at later time when intervention is considered),
  - **Catheter angiography** - confirmatory study and therapeutic tools in AVM. (shows feeding artery, nidus, and early draining veins).

Multidisciplinary Team



Indications for treatment

- **Location** – airway, eyes, mouth, ear,
- **Symptoms** – pain, functional impairment, cosmetic deformity
- **LM** - Hemorrhage, lymph leak , recurrent sepsis (local or regional)
- **VM** – chronic venous insufficiency, ulceration Thromboembolism
- **AVM** - High output heart failure, ischemic complications
- **Truncular lesions** can be handled conservatively until they become hemodynamically significant.

Options of treatment

General Measures

- Patient education and support
- Stop triggers
- Treat associated conditions
- Supportive measures
- Treatment plan
- Genetic and family member screening

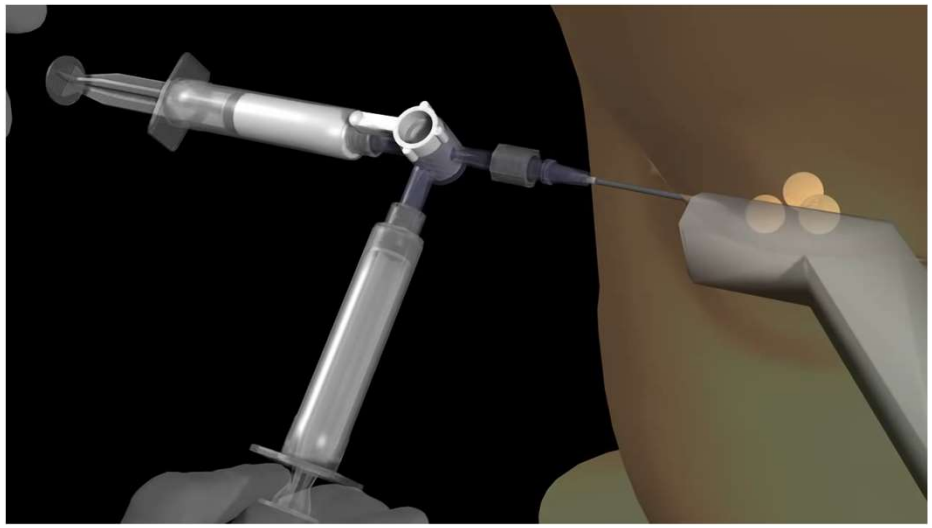
Conservative/Medical Therapy

- **Observation and monitoring** . (serial photo/imaging)
- **Precautions** - **AVM** (estrogen-containing contraceptives).
  LM (good skin care) **VM** (thromboembolism prophylaxis)
- **Compression therapy** (elastic garment and/or bandage)
- **Medication**
  - Thalidomide – reduce pain and bleeding in AVM
  - Sildenafil – mixed result in LM
  - Sirolimus (Rapamyin) - most promising, mainly in LM

Endovascular Therapy

- For surgically inaccessible extratruncular lesions,
- **Sclerothrapy**
  - ethanol, doxycycline, bleomycin, STS, OK-432
  - direct endothelial damage, thrombosis, and scarring
  - Have fever and alarming swelling afterwards
  - Complications: nerve injury, skin necrosis and cardiotoxicity
- **Embolization** (for high flow)
- **Other** – endovascular laser and radiofrequency ablation

Sclerotherapy



- **Play Video =>** Sclerotherapy: Adjunctive therapy pre- and postop. GA--localize cyst--aspirate--inject meds- 4-6wk apart --?4x

Surgery

- Resection/Excision = the only option for complete removal
  - requires extensive surgery with high morbidity.
  - Incomplete resection can result in recurrence, aggravation of lesion and interruption of endovascular access route.
- Other
  - procedures for AVM = Graft, ligation…
  - for lymphedema= debulking, microvascular anastomisis
  - For 2ry changes = Orthopedic and orthognatic procedures

Timing of treatment

- VM without bone involvement can usually be monitored until age 6 years. AVMs are also dormant for decades.
- Early intervention if limb or life-threatening area;
- Facial vascular malformation may benefit from early surgery.
- As most LM have comp. delay in their treatment is not indicated. When to perform surgical excision/sclerosis is unsolved.

Follow up

- Long term follow up required
- including radiographic studies and various noninvasive studies
- Female patients have much higher risks of deterioration or revival of extratrunclar lesion (menarch, pregnancy, hormone)
- Malformations affecting the extremity should be observed during the child's growth until the epiphyseal plate has closed

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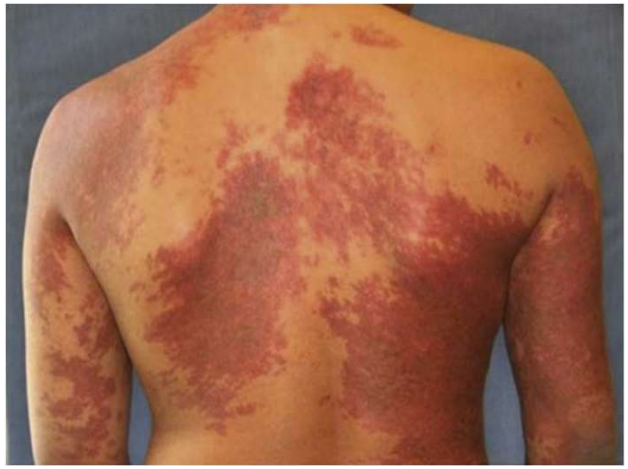
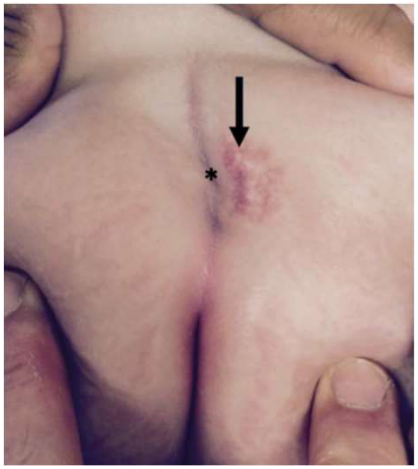
Capillary Malformations





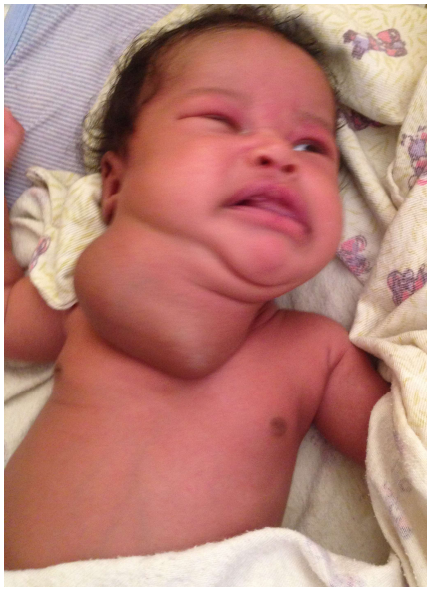
“port wine stain”. **Permanent**, pink-red, flat lesions. But nodules can develop overtime. **Pulsed dye laser therapy** will improve appearance On face -- warning for intracranial CVM. On spine--spinal dysraphism.

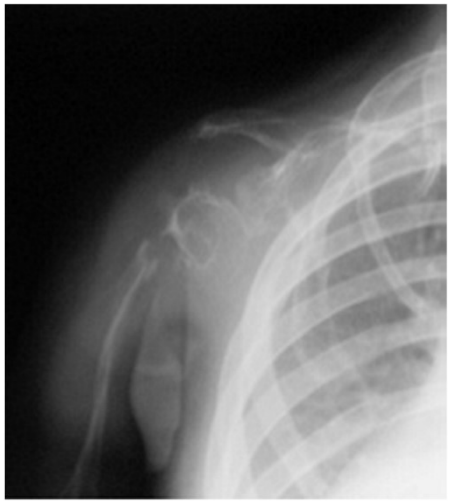
Lymphatic Malformations

Lymphatic Malformations

- Variously dilated lymphatic channels or **cysts**, lined with endothelial cells with a lymphatic phenotype (localized/diffuse infiltration)
- 50% at birth, 90% upto 2 yr
- can affect any site with the exception of the brain
- Cosmetic, mass effect – airway, dysphagia, speech,
- Comp = Hemorrhage (35%), infection (15-20%), and swelling
- Classification system more important than staging system (neck)

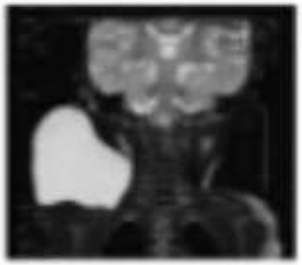
Lymphatic Malformations



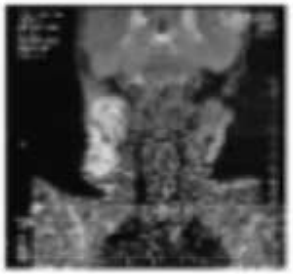


Soft compressible masses. usually don't involve the skin, can transilumiate if don't have septation. Cervical and axillary can extend to mediastinum. Can involve bone and skin (lymphangeiectasia)

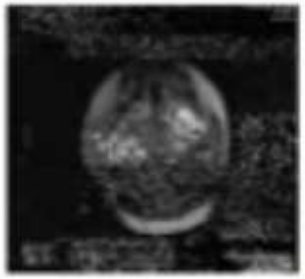
LM: Classification



Cystic Spaces > 2 cc
Historic "cystic hygroma"



>50% macrocystic



Cystic spaces < 2cc
Historic "lymphangioma"

There is no uniform consensus the definition. useful distinction is whether the cysts can be successfully aspirated/sclerosed,

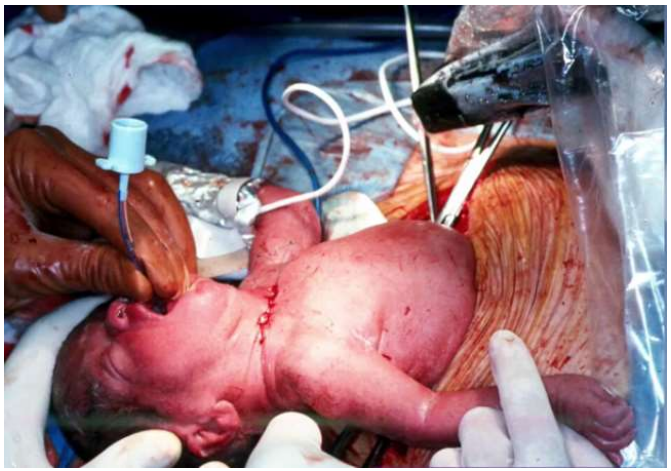
Imaging of LM vs VM

- **Duplex**
  - thicker walls vs thin septae
  - compressibility.
  - Flow larger patent VMs would demonstrate flow induced by compression
- **CT with contrast**
  - Enhancement of the vascular space,
  - presence of phleboliths and accompanying venous channels

LM: Natural history

- **Observation-** usually unacceptable by parents for large lesions
  - Spontaneous involution is rare (6-12%). More likely in macrocystic. Most likely after URTI
- **Enlargement in size** (Hemorrhage, infection)
  - ?start antibiotic no matter the cause,
  - sometimes aspirate to relive pressure
  - pain medication, rest, compressions

LM: Treatment

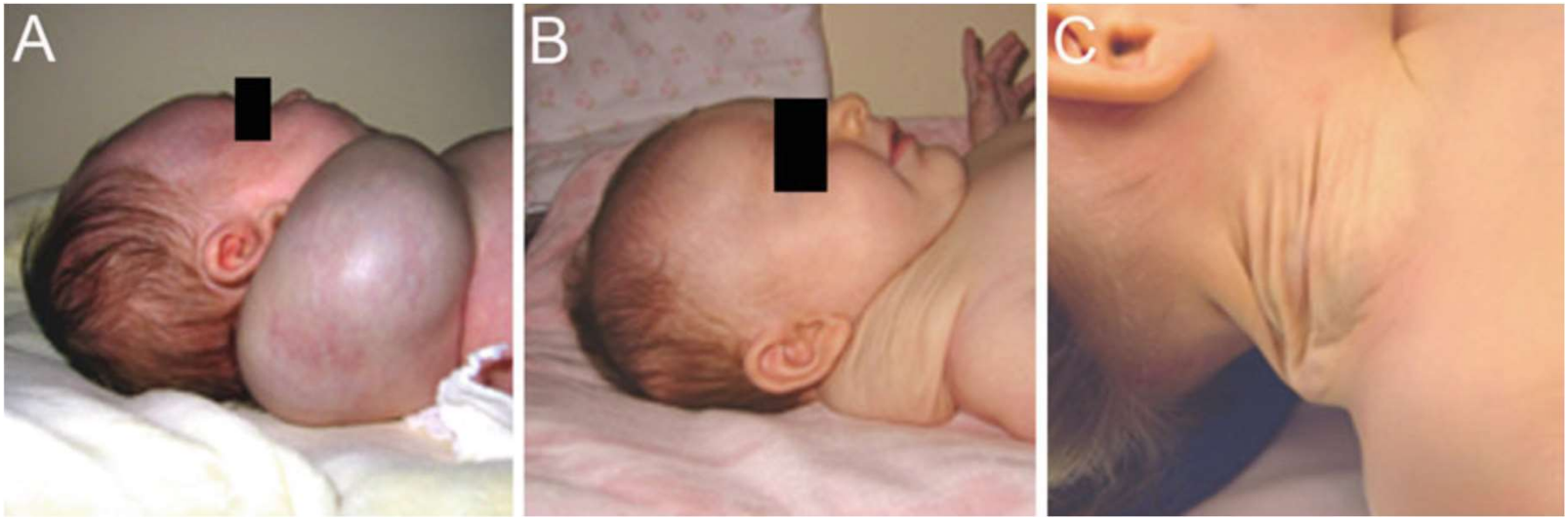


LM are the malformations needing treatment most frequently at very early age. Prenatal treatment is sometimes indicated, **EXIT** at delivery. **Bronchoscopy** is indicated followed by excision or tracheostomy.

LM: Treatment

- **Sclerothrapy** – surgically challenging areas. Mixed response to mixed, Poor response to microcystic. – observation/surgery
- **Surgical excision**
  - difficult to remove completely (vital structure and nerve)
  - Complete excision in 18-50%, **recurrence** rate of up to 53%
  - **complications** rate of 19–33%, **Bleeding** (most common) **nerve injury** 33%, **mortality** rate of 6%, and **extensive scarring**.

LM: Sclerotherapy

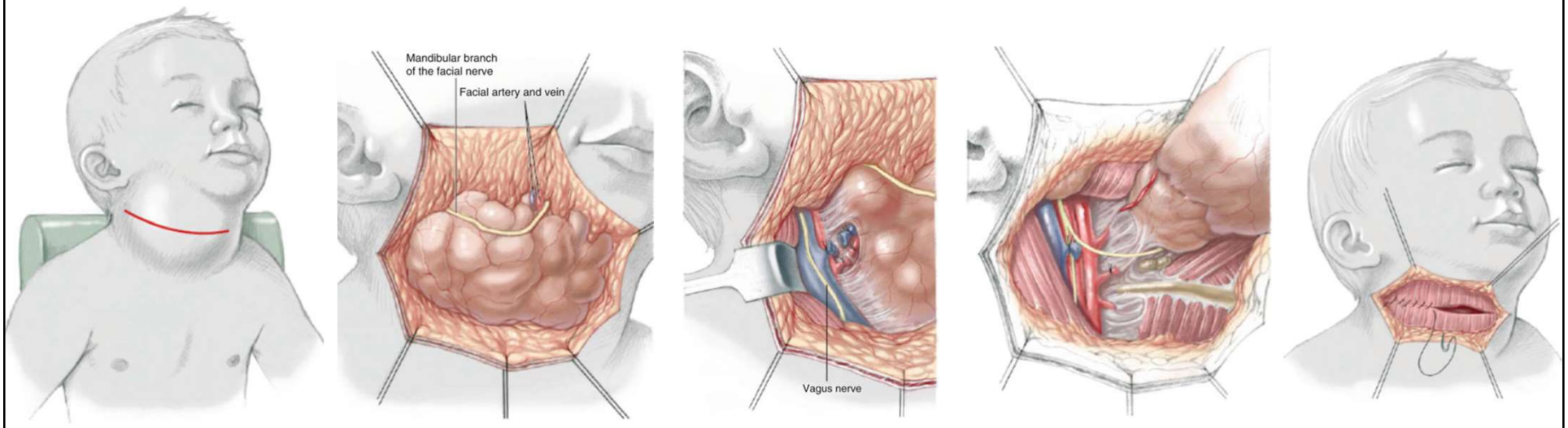


Sclerotherapy is ideally suited for **superficial macrocystic lesions**

LM: Surgery

- operations can be long and tedious, and often require meticulous dissection to preserve vital structures.
- **focus on a defined anatomic region**, removing as much of the lesion as possible without injuring vital structures;
- **limit blood loss** to less than the patient's blood volume;
- **prolonged closed suction drainage** of the resulting cavity is important.
- ? Sclerotherapy of the residual cavity following excision may be helpful in this regard

LM: Surgery



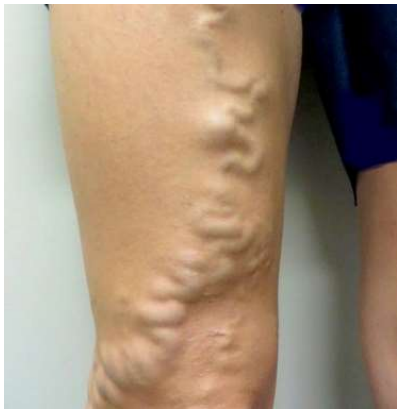
Large microcystic lesions in the neck are the most difficult benign lesions to resect. Raise subplatysmal flaps unless dermis is infiltrated. Expose facial N&V (superior), carotid, hypoglossal & vagus (medial). Then free mass from hyoid and SM gland

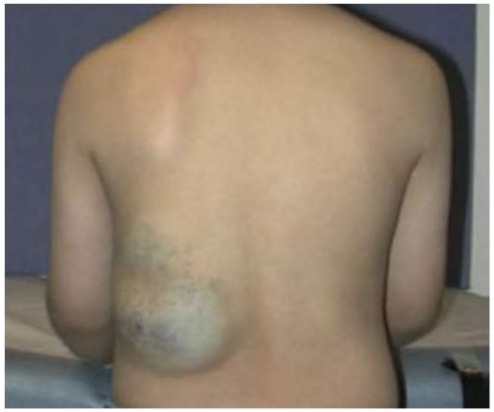
Venous Malformations

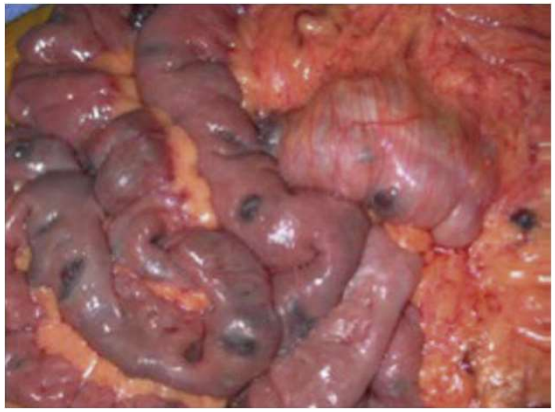
Venous Malformations

- **most common** symptomatic vascular malformation (2/3)
- 90% sporadic, 50% mutation in TIE-2 (*TEK gene*)
- **variable communication** to adjacent veins. The dilated venous channels are **thin walled**, compared with normal veins, and they usually **lack of venous valve**.
- Tend to form **thrombi**. Intralesional thrombosis can result in further expansion and deformity

Venous Malformations







"cavernous hemangiomas" range from simple **varicosities**, discreet spongy **masses**. Soft bluish swellings, can develop anywhere in the body, most commonly in the skin and soft tissues

VM: Coagulopathy

- Large VMs can be complicated by **localized** **intravascular coagulopathy** caused by stasis and stagnation of blood within the malformation, leading to consumption of coagulation factors.
    - **prolonged PT**, PTT normal
    - **decreased fibrinogen**, and **elevated D-dimers**.
    - **Thrombocytopenia** can occur <100,000/µL.
- Prevention with **low-dose aspirin** may be beneficial

VM: Treatment

- **Compressive therapy:** usually Class II (20– 30 mmHg) is adequate
- **Intralesional sclerotherapy** is the mainstay of treatment
  - injected under **fluoroscopy**,
  - with the use of **tourniquets and compression** of venous drainage to prevent systemic administration
- **Resection** is typically reserved for well localized lesions, but is marked by procedural morbidity and recurrence,

OUTLINE

1. Embryology
2. Classification
3. Diagnostic approach
4. **Low flow lesions**
5. High flow lesions

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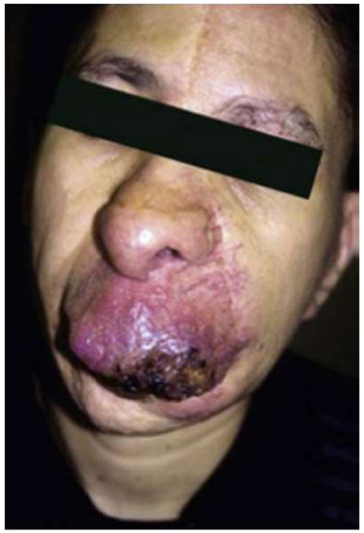
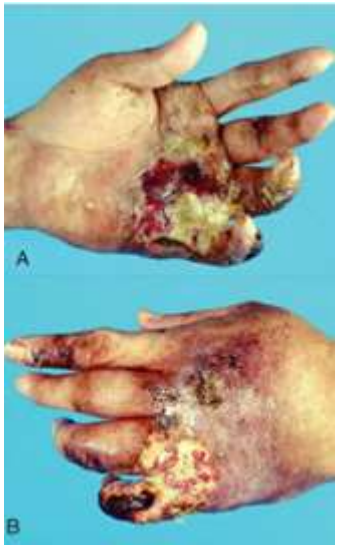
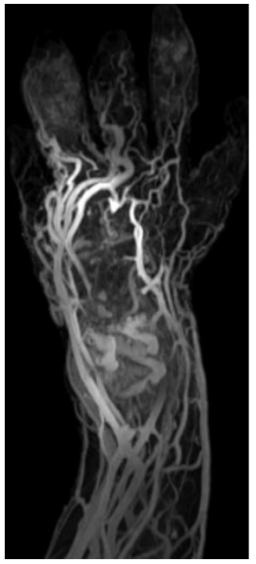
Areteriovenous Malformations

- **Abnormal connections** or **shunts** between feeding arteries and draining veins, without an intervening capillary bed
- **CNS** is more frequent than extracranial AVM.
- At **birth**, they appear as a **pink cutaneous blemish** that can be confused with a CM.
- fast flow through the shunt becomes more evident in childhood and **adolescence** as the lesion expands and develops into a **mass**
- Fortunately, AVMs are the least common but the most aggressive and the most difficult to treat.

Areteriovenous Malformations







Skin ischemia can develop from expansion or local steal phenomenon, leading to pain, ulceration, and bleeding

AVM: Treatment

- **Angiographic embolization** alone or in combination with excision (2-3d later)
- **Direct puncture sclerotherapy** of the AVM nidus can be an adjunct to embolization,
- **Surgery** - Both the nidus of the AVM and the involved skin should be removed. Observation of the pattern of bleeding at the resection margins and frozen section can guide extent..
- For difficult AVMs of the extremities (distal), **amputation** becomes an option.

OUTLINE

1. Embryology
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THANK YOU!

References

- **Hollcomb and Aschcraft pediatric surgery** 7th edition, 2020
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