Pediatric Vascular Disorders

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Disclosures

- I have no financial relationships to disclose.
Vascular Tumor vs Malformations

- **Vascular tumors** are true cellular proliferations of endothelial cells
  - Includes infantile hemangioma, later onset pyogenic granuloma
  - Less commonly: tufted angioma, kaposiform hemangioendothelioma

- **Vascular malformations** are due to defects in morphogenesis
  - True angiogenesis may occur leading to expansion and thickening
  - Subcategorized as fast flow or slow flow
    - **Slow flow**: capillary, venous, and lymphatic malformations
    - **Fast flow**: arteriovenous malformations

VASCULAR TUMORS
Infantile Hemangiomas

- Most common tumors in the neonatal period, 4-5% of infants, most often noted in the first several weeks of life
- Significant growth over the first several months*
- Spontaneous involution over the years – note: may be incomplete, may leave scarring* - Involution distinguishes these lesions from malformations
- Risk factors include: female, Caucasian, low birthweight, premature, multiple gestation, older mothers
- Margileth and Museles reported a 10% familial incidence
Pathogenesis – Defects in Signaling Theory

- Several hypotheses with no single theory explaining all features

- Somatic mutations in genes involved in the VEGF signaling pathway
  - Shift from VEGFR2 $\rightarrow$ VEGFR1

- Germline mutations in VEGFR2 and TEM8 found in a small subset

- Familial cases linked to chromosome 5q – suggests involvement in genes at this locus

Pathogenesis – Placental Hypothesis and GLUT-1 Theory

- **Placental** hypothesis
  - Hemangiomas share an immunohistochemical phenotype with placental cells
  - Suggesting that hemangiomas are:
    - 1) are of placental origin, via embolization - or –
    - 2) undergo differentiation toward a placental microvascular phenotype

- **GLUT-1**
  - Expressed by infantile hemangiomas and placenta, not by other vascular tumors or malformations
  - Other vascular antigens expressed by hemangiomas include: merosin, Fc gamma RII, and Lewis Y antigen
Pathogenesis – Hypoxia Theory

- Supported by the occurrence of hemangiomas with hypoxic placental changes, prematurity, low birthweight, retinopathy of prematurity, and regional arterial insufficiency syndromes e.g. PHACE(S)

- Hypoxia $\rightarrow$ upregulates GLUT-1 and VEGF $\rightarrow$ mobilization of endothelial progenitor cells

- Hypoxia and estrogen have a synergistic effect on endothelial cell proliferation in vitro
Pathogenesis of the Transition From: Proliferation to Involution

- **Indoleamine 2,3-dioxygenase (IDO)** degrades tryptophan
  - Starves T cells of tryptophan → inhibits T cell activation
  - Believed to protect the fetus from rejection, and as a secondary effect, protects proliferating hemangiomas from immune surveillance
  - IDO highly expressed by macrophages, dendritic cells, and placenta
  - **Downregulation of IDO** may lead to involution

- **DUSP5** involved in apoptosis; somatic mutations in this gene may play a role in proliferation and involution
Clinical Presentation

- **Superficial hemangiomas** in the superficial dermis (50-60%)
  - Bright red in color during the proliferative phase
  - Finely lobulated surface, ‘strawberry hemangioma’
  - Often focal, but may be of the larger plaque type or segmental type (may have extracutaneous involvement e.g. PHACE(S))

- **Deep hemangiomas** in the deep dermis and/or subcutis (25-35%)
  - May not be evident in the first few weeks
  - Warm, ill-defined, blue to purple masses with no to minimal overlying changes
  - Often have a significant arterial supply and a bruit may be felt

- **Mixed** (15%)

- 25% have multiple lesions and may have visceral involvement

Natural History

Proliferative Phase:

- Maximal radial growth often established early on
- **80% achieve final size by 3.2 months**, on average
- Become warmer and finer in texture, surface may appear tense
- **Deeper lesions tend to proliferate for a month longer** than superficial lesions
- Deeper component may continue to proliferate after the superficial component has plateaued
- Small subset of lesions without a proliferative phase

Involution:

- Begins in the first year and persists for several years
- Changes from bright to a gray-purple with flattening, assumes fatty consistency
- **30% by 3 years, 50% by 5 years, 70% by 7 years, and 90% by 9 years**
- May leave atrophic, fibrofatty, or telangiectatic changes

Complications

- **Ulceration**
  - Can occur in **up to 10%**, on average at 4 months
  - Whitish discoloration may be a sign of impending ulceration
  - High risk areas include the **lips, anogenital region, and skin folds (neck)**
  - More common with large, mixed, and segmental types
  - Those with **minimal to no growth phase are at higher risk**
  - Bleeding is a rarely significant and can be managed with firm pressure

- **Kasabach Merritt Phenomenon** – thrombocytopenic coagulopathy
  - Associated with **kaposiform hemangioendotheliomas** and **tufted angiomas**
  - Presents within the first few months to a year of life
  - Tumor becomes indurated, enlarged, with local petechiae and purpura

Complications

- **Periocular** lesions
  - Can compress the lobe leading to an **astigmatism**
  - May obstruct vision and invade the orbital musculature → **amblyopia and strabismus**
  - **Proptosis** is a rare presentation of an orbital hemangioma; Evaluation by ophthalmologist

- **Nasal tip** – may distort the underlying cartilage

- **Lip** – painful ulceration is common, may interfere with feeding

- **Pinna** – may ulcerate and become infected, conductive hearing loss

- **Breast** – may affect the underlying breast bud and lead to breast asymmetry

- **Anogenital** – painful defecation

Systemic Involvement

- Large segmental lesions often associated with extracutaneous anomalies

- Cerebrovascular (91%), cardiovascular (67%), structural brain (52%) anomalies associated with PHACE(S) syndrome

- ‘Beard region’ – possible laryngeal hemangiomatosis
  - Airway hemangiomas can be present, often in the subglottic area
  - May present as noisy breathing, biphasic stridor
  - Referral to ENT

- Midline lumbosacral region
  - Occult spinal dysraphism in 35% of lesions >2.5cm
  - Other risk factors: ulceration, deviated gluteal cleft, lipoma, skin appendages

- Large hemangiomas of the lower body may be seen in LUMBAR syndrome

Visceral Involvement

- If 5 or greater lesions, recommend evaluation for visceral involvement

- Liver most commonly involved
  - Complications include high output cardiac failure, abdominal compartment syndrome, and hypothyroidism
  - Hemangiomas may produce type 3 iodothyronine deiodinase which deactivates thyroid hormone, most often associated with hepatic hemangiomas
    - Resolves with regression of lesion
    - Consider serial ultrasounds, lab evaluation for hypothyroidism, cardiac evaluation

- GI tract involvement → bleeding

- CNS hemangiomas are rare (1%), may present with hydrocephalus
Evaluation

- **MRI, +/- contrast** is best for evaluating the extent and tissue characteristics

- If equivocal, or concerns for malignancy, **consider a biopsy**

**Pathology – Infantile hemangiomas**

- **Proliferating** phase: non encapsulated masses composed of proliferating plump endothelial cell and pericytes
- **Late proliferating** phase: lobules of endothelial masses separated by fibrous septae with larger vessels within the septae
  - Mitotic figures and apoptotic bodies may be present
  - Increased number of mast cells
- **Involuting** phase: flattening of the endothelium, reduced mitotic figures, decrease in # of vessels with fibro fatty tissue, reduced # mast cells

**Pathology - Congenital hemangiomas**

- Striking lobularity with densely fibrotic stroma, stromal hemosiderin deposits, focal thrombosis and sclerosis of capillary lobules, lack of infiltration to tissues, fewer mast cells, proliferating thin walled vasculature with multiple thin walled vessels
- May demonstrate zones of involution

EVALUATION OF A CHILD WITH INFANTILE HEMANGIOMAS FOR POSSIBLE SYSTEMIC INvolvement

**Multiple (≥5) hemangiomas***
- Abdominal (hepatic) ultrasound
- CBC, stools guaiac; consider urinalysis

**Beard** hemangiomas
- ENT evaluation, laryngoscopy
- Exclude PHACE(S) syndrome

Segmental hemangioma
- Cervicofacial**: exclude LUMBAR Syndrome

**Initial evaluation** (preferably at age <3 months)
- Physical examination of affected region
- Ultrasound (with Doppler) of abdomen, pelvis and (if age <3 months) spine

**Additional evaluation** (preferably at age 3-6 months, unless otherwise indicated)
- If midline lumbar hemangioma or lipoma (or abnormal spinal ultrasound):
  - MRI of spine and consider neurosurgical evaluation
  - If extensive diaper area involvement (or abnormal pelvic/renal ultrasound):
    - Consider urologic evaluation
    - If extensive limb involvement: orthopedic evaluation and additional imaging as clinically indicated

- CT, computed tomography; MRI: magnetic resonance imaging; MRA: magnetic resonance angiography; MRV: magnetic resonance venography; TIIIs: thyroid function tests

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* In infants ≥6 months of age and as clinically indicated in older infants.

** Consider evaluation for hepatic/gastrointestinal tract involvement and thyroid abnormalities if a large lesion.

† LUMBAR: lower body/lumbosacral hemangioma, lipoma/other skin lesions; urogenital anomalies; ulceration; myelopathy (spinal dysraphism); bony deformities; anorectal malformations; arterial anomalies; renal anomalies. Also referred to as SACRAL (spinal dysraphism, anogenital anomalies, cutaneous anomalies, renal/urologic anomalies, angioma, in lumbosacral area) and PELVIS (perineal hemangioma, external genital malformations, lipomyelomeningocele, vesicorenal abnormalities, imperforate anus, skin tag) syndrome.
RICH and NICH

- Congenital hemangiomas, fully developed at birth
- Distinct from infantile hemangiomas in that they are GLUT-1 negative
- Rapidly involuting congenital hemangioma (RICH)
  - Significant intrauterine growth followed by little to no growth postnatally
  - Rapid involution with the first year, may ulcerate, necrose, or bleed
  - May be associated with transient thrombocytopenia
- Non-involuting congenital hemangioma (NICH)
  - Unlike infantile hemangiomas, grows proportionately with the child
  - May worsen with maturity and do not spontaneously involute

RICH: Spontaneous involution at 5 months
Lesion in a school aged child with no change since birth

Treatment – Topical Therapies

- **Goals:** prevent/reverse life threatening complications, treat ulcerations, prevent disfigurement, avoid overly aggressive treatments that may lead to scarring

- **Non intervention**
  - Appropriate for **small hemangiomas**
  - Important to discuss care for superficial bleeding and ulceration
  - Ulceration tx: local wound care, pulsed dye laser, pain control

- **Local therapies**
  - **IL corticosteroids, TAC 5-40 mg/mL** have been used successfully
    - Caution in periorbital area, consider potent class I topical steroid
  - **Beta blockers** (e.g. topical timolol 0.5% gel, topical imiquimod)

*4 weeks of topical timolol gel, 1 drop BID*
Treatment – Systemic Therapies

- Systemic Corticosteroids
  - Individualize dosage and tapering regimen
  - Prednisone: 2-3 mg/kg/day to 3-5 mg/kg/day
  - Side effects: cushingoid facies, irritability, disruption of sleep, GI sx, decreased growth rate (with catch up), HPA axis suppression, immunosuppression
  - Live vaccines not recommended
  - Bactrim for PCP prophylaxis

- Systemic Beta Blockers***
  - Binds B_2_ adrenergic-R on hemangioma endothelial cell → vasoconstriction and decreased expression of VEGF, bFGF → apoptosis
  - No consensus on dosage, 2-3 mg/kg used in studies, taper to prevent rebound tachycardia
  - Side effects: hypotension, bradycardia, hypoglycemia, bronchospasm, sleep disturbances
  - Baseline cardiac evaluation and pediatric cardiologist referral prior to therapy

- Pulsed Dye Laser
  - Consider for superficial hemangiomas, not effective for deeper lesions
  - May result in pigmentary alteration, atrophic scarring, and ulceration

- Surgical Excision
  - Best reserved for involuted lesions to remove fibro fatty tissue and redundant skin
  - Also consider in cosmetically sensitive areas such as the nasal tip or lip
PHACES

- Posterior fossa malformations
  - Dandy-Walker is the most common

- Hemangioma of the face
  - Usually plaque-like, more than one dermatome

- Arterial abnormalities

- Coarctation of the aorta

- Eye abnormalities

- Sternal nonunion

- Supraumbilical raphe
PHACES

Diffuse Neonatal Hemangiomatosis

- Cutaneous and visceral hemangiomas
- Liver hemangioma may be complicated by obstructive jaundice
- Prognosis depends on which organ systems are involved
- If the patient has multiple cutaneous hemangiomas: US, UA, stool guaiac, CBC
- Mortality in severely affected infants may occur secondary to liver disease, GI bleeding, neurological problems, high output cardiac failure and respiratory compromise
- If no visceral involvement: benign neonatal hemangiomatosis
Diffuse Neonatal Hemangiomatosis

Tufted Angioma

- Onset during infancy or early childhood
- Presents as ill-defined red-brown plaque or patch over the neck or upper trunk
- Plaque slowly extends over time
  - Typically does not regress
Tufted Angioma

PELVIS Syndrome

- Perineal hemangioma
- External genital malformations
- Lipomyelomeningocele
- Vesicorenal abnormalities
- Imperforate anus
- Skin tags
Pelvis Syndrome

Pyogenic Granulomas

- Presents as rapidly growing, friable skin or mucosa with frequent ulceration
- Common in children and young adults
- Associated with antecedent drama, pregnancy, oral medications (retinoids, inidinavir, and EGFR inhibitors)
Pyogenic Granulomases

Kaposiform Hemangioendothelioma

- **Characteristics**
  - Rare; usually occurring in children younger than 2
  - Male-female incidence equal
  - Presents as a rapidly growing vascular macule, plaque, nodule, or bulging indurated mass
  - Associated with *Kasabach-Merritt phenomenon*: life-threatening thrombocytopenia from platelet trapping within the tumor; likely caused by retroperitoneal tumors

- **Pathology**
  - Densely infiltrated nodules composed of spindle cells with minimal atypia and slit-like vessels containing hemosiderin; GLUT-1 negative

- **Treatment**
  - Corticosteroids are first-line; complete surgical excision if feasible; vincristine is the first-line treatment for Kasabach-Merritt phenomenon
Kaposiform Hemangioendothelioma

Glomeruloid Hemangioma

- **Characteristics**
  - A type of hemangioma seen in **POEMS** (polyneuropathy, organomegaly, endocrinopathy, monoclonal gammopathy, skin lesions) syndrome
  - Firm, red-to-purple papules on the trunk or the extremities

- **Pathology**
  - Appears similar to a renal glomeruli; dilated dermal vessels filled by small, well-formed capillary loops

- **Treatment**
  - Not required, but consider shave excision, cryosurgery, electrodessication, or pulsed dye laser surgery
Glomeruloid Hemangioma in POEMS Syndrome

http://www.pathologyoutlines.com/topic/skintumor nondermalepitheliomaepidermolysis.html
VASCULAR MALFORMATIONS
Vascular Malformations

- **Capillary malformation**: a group of disorders that developed from changes in the blood/lymphatic channel formation

- **Venous malformation (VM)**: recognized clinically as a well-demarcated patch or plaque with a blue hue with compressibility and ability to refill with dependency
  - Can be focal, segmental, or widespread
  - T2-weighted MRI is the best imaging modality
  - Cephalic VM: develop into cosmetic and function problems with time → distortion of facial features, sleep apnea (laryngeal VM), skull defects
  - Trunk and limb VM: spongy masses that can be emptied with elevation and massage; can involve muscles and joints
Vascular Malformations

- **Lymphatic malformation**: an abnormal proliferation of lymphatic channels

- Vascular malformations are further classified as “fast-flow” and “slow-flow”
  - Fast-flow: arteriovenous malformations (AVM)
  - Slow-flow: Capillary, venous and lymphatic malformations
Capillary Malformations
Port-Wine Stain

- Port-Wine Stains (PWS) are capillary malformations that are usually present at birth.
- Can be localized, segmental, or multifocal.
- PWS tend to grow with the child and do not regress.
- Facial PWSs are commonly located along the sensory trigeminal nerve: V1 (ophthalmic), V2 (maxillary), and V3 (mandibular).
- As the patient ages, PWSs tend to become darker and more violaceous, especially when located on facial V2-V3 distributions.

**Nevus Simplex and Salmon Patches are NOT PWSs, rather congenital capillary stains.**
Nevus Simplex

Photo courtesy of Neonatal and Infant Dermatology, 3rd edition. Eichenfield, Frieden, Mathes & Zaenglein
Port-Wine Stain

Port-Wine Stain

This lesion involves both the V1 and V2 trigeminal dermatomes in this infant with Sturge–Weber syndrome.

Phakomatosis Pigmentovascularis (PPV)

The combination of a capillary malformation (Port-Wine Stain) with different melanocytic neoplasms

Types:

- Ia/b: PWS + epidermal nevus
- IIa/b (Phakomatosis cesioflammea): PWS + Mongolian spot +/- nevus anemicus
- IIIa/b (Phakomatosis spilorosea): PWS + nevus spilus +/- nevus anemicus
- IVa/b (Phakomatosis pigmentovascularis): features of II + III or II + V
- Va/b (Phakomatosis cesiomarmorata): Mongolian spot + cutis marmorata telangiectatica congenita
Phakomatosis Pigmentovascularis

Photo courtesy of Neonatal and Infant Dermatology, 3rd edition. Eichenfie1, Frieden, Mathes & Zaenglein
Sturge-Weber Syndrome (Encephalotrigeminal Angiomatosis)

- Sporadic neuroectodermal syndrome
- Characterized by
  - Port-wine stain at birth in trigeminal distribution
    - Greater risk of SWS – if PWS involves all (V1,V2,V3) branches, bilateral, both upper and lower eyelids
  - Leptomeningeal angiomatosis (ipsilateral to PWS)
    - Resultant seizures
  - Ocular involvement – 60% of SWS patients
    - Glaucoma most common, ocular AVM’s, nevus of Ota, buphthalmos, and blindness.
- Other features
  - Mental retardation, cerebral atrophy, tram-track cortical calcifications, enlarged choroid plexus
Sturge Weber

Photo courtesy of *Neonatal and Infant Dermatology, 3rd edition*. Eichenfield, Frieden, Mathes & Zaenglein.
Klippel-Trenaunay Syndrome

- Sporadic disorder
  - Triad of Vascular malformations, venous varicosity, hyperplasia of soft tissue and bone.
  - MC vascular malformation is PWS (LE > UE)
  - Lesions of KT are usually all on same extremity

- Parkes-Weber syndrome –
  - Includes Arteriovenous malformations.
Klippel-Trenaunay Syndrome

This patient has the characteristic triad, including port-wine stain, hemihypertrophy, and venous varicosities.
Telangiectasias (Spider & Angioma Serpiginosum)

- Not true capillary malformations.
- May appear in infancy or early childhood.
- They are usually composed of small, punctate linear vessels distributed in either a segmental, unilateral nevoid, or diffuse pattern.
- Children sometimes develop so-called spider angiomas comprised of a brightly erythematous central punctum with radiating ‘spider-like’ telangiectasias. They are usually located on sun-exposed areas.
- Risk factors include fair skin and a history of minor skin injury at the site.
- These may disappear spontaneously or persist throughout life.
Spider Angioma

Photo courtesy of *Neonatal and Infant Dermatology, 3rd edition*. Eichenfield, Frieden, Mathes & Zaenglein.
Telangiectasia (Spider & Angioma Serpigenosum)

- **Angioma serpigenosum:** Clusters of dark red puncta in a serpiginous pattern. Typically on an extremity and typically female.

Photo courtesy of *Neonatal and Infant Dermatology, 3rd edition. Eichenfield, Frieden, Mathes & Zaenglein.*
Cutis Marmorata Telangiectatica Congenita (CMTC)

- Dark red to purple reticulated vascular pattern that is broad
- Usually affects one or more limbs and the trunk
- Often associated with telangiectasias +/- prominent veins
- Persists upon warming
- Often lightens in color over the first year of life
CMTC

Neonate with CMTC (Photos one year apart)

Photos courtesy of Neonatal and Infant Dermatology, 3rd edition. Eichenfield, Frieden, Mathes & Zaenglein
Hereditary Hemorrhagic Telangiectasia (HHT)

- AKA: Osler-Weber-Rendu disease
- AD; ENG (endoglin) and ALK1 (activin receptor-like kinase 1) genes
- Multiple telangiectasias on mucosal and cutaneous surfaces.
- First manifestation: nose bleeds in children
- Telangiectasias typically appear in adolescence and adulthood.
- Internal organs may be affected and screening for pulmonary and CNS AVMs is imperative
Ataxia-Telangiectasia (Louis-Bar Syndrome)

- Multi system autosomal recessive disorder
- ATM gene defect (chromosome 11q)
  - Mild form ATM – due to MREII gene defect
- High rate of chromosomal breakage and sensitivity to ionizing radiation

Characterized by
- Truncal ataxia – as early as infancy
  - Other neurologic features include choreoathetosis, dysarthria, myoclonic jerks, oculomotor abnormalities.
- Oculocutaneous telangiectasias – around 3-5 years old
  - Medial/lateral bulbar conjunctivae, and other sun exposed sites
- Profound humoral/cellular immunodeficiency
  - IgA, IgE, IgG decreased with chronic sinopulmonary infections
- Other features
  - Predisposition to hematologic malignancy, skin cancers, subcutaneous fat loss, premature aging of skin and hair, Non-infectious cutaneous granulomas
Ataxia-Telangiectasia

Angiokeratoma (5 types)

- Angiokeratomas are essentially telangiectases that have an overlying *hyperkeratotic* surface.

- 5 types:
  - Angiokeratoma circumscriptum
  - Angiokeratoma corporis diffusum
  - Angiokeratoma of Mibelli
  - Angiokeratoma of the scrotum (Fordyce)
  - Solitary angiokeratoderma
Angiokeratoma

Angiokeratoma

- **Angiokeratoma circumscriptum:**
  - Malformation of dermal and subcutaneous capillaries and veins, and is variably classified as a capillary or venous malformation. The vascular malformation is congenital.
  - Superficial ablative therapy is typically followed by recurrence, regardless of whether ablation is performed by excision, laser, cryotherapy, or electrocautery.
Angiokeratoma

- **Angiokeratoma corporis diffusum – AKA Fabry Disease:**
  - Fabry disease is a rare X-linked lysosomal storage disease. It is caused by mutations in the alpha-galactosidase A gene (GLA), leading to a deficiency in alpha-galactosidase A.
  - Angiokeratomas occur in 66% of males and 36% of females with Fabry disease. The average age of onset in males is about age 20; in females it is about 10 years later.
  - Lesions tend to occur in the “bathing trunk” area, from the umbilicus to the genitalia.
  - Telangiectasias occur in about 25% of male patients presenting around age 25 and in women around age 40. The vascular lesions can be treated with intense pulse light or various vascular lasers.
Fabry Disease

Angiokeratoma

- **Angiokeratoma of Mibelli:**
  - Usually appear as 1–5 mm red vascular papules, which become hyperkeratotic over time.
  - The papules are dull red or purplish-black, verrucous, and rounded, and are usually situated on the dorsum of the fingers and toes, the elbows, and the knees. Frequently, these are called telangiectatic warts.
  - The patient often has cold, cyanotic hands and feet.
  - This is a rare genodermatosis with an autosomal-dominant trait for vascular lesions located over bony prominences and a family history of chilblains. The condition is most frequently discovered in prepubertal children.
  - Treatment includes electrocautery, fulguration, CO₂ laser ablation, long-pulse vascular laser therapy, or cryotherapy, with fairly good results.
Angiokeratoma

- **Fordyce Angiokeratoderma:**
  - Multiple small vascular papules that stud the scrotum and sometimes the vulva in middle-aged and elderly individuals.
  - There is often a diffuse redness of the involved area that may be a source of concern to the patient. Urethral or clitoral lesions may also be seen.
  - Treatment is best accomplished by shave excision, cautery, laser ablation, or fulguration of troublesome lesions. The primary therapy is reassurance.
Fordyce Angiokeratoma

Angiokeratoma

- **Solitary Angiokeratoderma:**
  - A single small, bluish-black, warty papule that occurs predominantly on the lower extremities.
  - It is not a hereditary lesion and probably follows trauma, with subsequent telangiectasia before the formation of the angiokeratoma.
  - The mode of acquiring this lesion, and its small size, solitary nature, and location, distinguish it from other forms of angiokeratoma.
  - Solitary angiokeratoma is to be considered in the differential diagnosis of seborrheic keratosis, melanoma, pigmented basal cell carcinoma, and ordinary hemangioma.
  - Treatment is by electrosurgery, laser ablation, or excision.
Venous Malformations
Venous Malformations

- MRI to determine extent of involvement
- Localized intravascular coagulopathy and treat accordingly
- Manage site-specific complications

Courtesy of Bolognia, J., Jorizzo, J. L., & Schaffer, J. V. (2012). Dermatology
Venous Malformation

Small venous malformation of the finger
Syndromes Associated with VMs

- **Familial cutaneous and mucosal venous malformation (VMCM)**
  - Autosomal dominant condition with VMs affecting skin, oral mucosa, and muscles; some visceral VM (intestines, lungs, CNS) noted
  - Mutation in the *TEK* gene

- **Blue rubber bleb nevus syndrome (BRBNS)**
  - Sporadic disease with dark blue papules and compressible protuberances
  - Gastrointestinal involvement can lead to bleeding and iron deficiency anemia

- **Glomuvenous malformation (GVM)**
  - Variant of VM with rows of glomus cells around venous channels
  - Solitary, blue-purple nodules usually without visceral involvement
  - Mutation in the glomulin (*GLMN*) gene

- **Maffucci Syndrome**
  - Sporadic condition with a combination of VMs and enchondromas most commonly on the extremities
  - PTHR1 gene
Syndromes Associated with VMS

Blue Rubber Bleb Nevus Syndrome

Glomuvenous Malformation

Maffucci Syndrome

Glomangioma

- Glomuvenous malformations (GVM; formerly called ‘glomangioma’) may be solitary or multiple, and may be localized or involve a larger territory of skin.

- Cutaneous Findings: They are often bluish to purple, cobblestoned or plaque-like in appearance.

- During childhood, they typically acquire a deeper blue hue and thicken, and become tender when palpated.

- Congenital plaque-like GVMs are usually pink at birth, with noticeable thickening and change in color to blue-purple during childhood.

- These plaque-like GVMs may arise sporadically, or occur as a manifestation of autosomal dominant GVM.
Glomangioma

- EXTRA CUTANEOUS: In addition to skin and mucosal involvement, VM can also involve deeper soft tissues, muscles, joints, and in severe cases, visceral sites such as the abdomen and pelvis.

- DIAGNOSIS: Usually established by clinical features, but imaging may be used as well - ultrasonography, Doppler, MRI, and CT scans are useful for evaluating the extent of involvement.

- TREATMENT: VMs can be treated with many different modalities including sclerotherapy, excision, endovenous laser ablation, cutaneous laser ablation or a combination of these modalities.

- Sclerotherapy has emerged as the mainstay of treatment for VMs, followed by surgical excision.

- The decision regarding when to treat is usually based on whether there is significant functional impairment or disfigurement.
Glomangioma

Photos courtesy of *Neonatal and Infant Dermatology, 3rd edition*. Eichenfield, Frieden, Mathes & Zaenglein.
Lymphatic Malformations
Lymphatic Malformations

- Hyperplasia of the lymphatic network
- Can be superficial (skin, mucous membranes), deep (bone, muscle), or visceral

Primary:
- Abnormal development of lymphatic system

Secondary:
- Abnormal distribution of lymphatic channels
Lymphangioma

- Lymphatic Malformation – aka Lymphangioma – are slow-flow vascular anomalies.

- 4 types:
  - Macrocystic
  - Microcystic
  - Combined
  - Generalized (rare)

- Etiology is unknown – cases are sporadic.
Lymphangioma

- Cutaneous Findings are visible at birth and can even be found on prenatal US.
- They occur commonly on the neck and axilla and can be referred to as **Cystic Hygroma**
- Extracutaneous finding include intraoral involvement, mandibular and orbital locations as well as visceral and abdominal lesions.
- Treatment for these benign lesions is typically directed at managing complications or restoring anatomy.
- Treatment modalities include:
  - Sclerotherapy, compression, surgical resection and RF ablation
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Resources


- Andrews’ Disease of the Skin, 11th edition. James, Berger, Elston