VASCULAR MALFORMATIONS

A. Common features

- Always present at birth (distinguishing factor from hemangiomas, which grow after birth and are not often seen at birth).
- Vessels are inherently abnormal due to aberrant signal pathways that determine apoptosis and proliferation pathways.
- 1:1 male: female ratio
- Lesions grow proportionately to child and do not involute.

B. Diagnosis

- Clinical history and physical examination
- Imaging
  a. US with Doppler
    i. Differentiates slow-flow from fast-flow lesions
    ii. Highly operator-dependent
  b. MRI with contrast
    i. Gold standard test
    ii. Provides details on the anatomic distribution of the lesion along with unique sequences and signals used to differentiate the types of vascular malformation
  c. Arteriography
    i. Invasive study
    ii. Most often used in conjunction with planned embolization

Capillary malformations

- Slow-flow lesion
- Appearance: Regular, dilated, thin-walled capillaries localized to the papillary and superficial reticular dermis.
  a. Must be differentiated from other common macular stains of the face (e.g., nevus flammeus).
  b. The autonomic nervous system influences the development of this lesion, which is why it is often localized to distinct nerve distributions (e.g., V1 nerve distribution with port-wine stains of the face).
- Incidence: 0.3% of newborns. Most common location is on the face
- 3:1 female: male ratio
- Common associated syndromes

1. Sturge–Weber syndrome
    i. Port-wine stain (capillary malformation) in V1/V2 distribution of the face
    ii. Leptomeningeal malformations
      a) Seizures
      b) Contralateral hemiplegia
      c) Warrants MRI of head

iii. Developmental delay
iv. Glaucoma and retinal detachment: Screen using biannual fundoscopic and tonometry examinations for 1 to 3 years, then yearly examinations

2. Klippel-Trenaunay syndrome
   i. Capillary malformation and/or lymphatic-venous malformation (patchy port-wine stain on an extremity)
   ii. Skeletal and soft tissue hypertrophy (axial/transverse) of an extremity
3. Parkes–Weber syndrome
   i. Capillary malformation + AVM
   ii. Soft tissue/skeletal hypertrophy
4. Cobb syndrome
   i. Capillary malformation localized to the trunk
   ii. Associated with spinal AVM

- Treatment options

a. Observation
   i. Lesions do not regress
   ii. Can progress to "cobblestone" appearance

b. Pulsed dye laser
   i. Typically requires multiple treatments
   ii. 70% to 80% of patients respond with decrease in pigmentation of the lesion.
   iii. More favorable results on the lateral face as compared to mid-face, trunk, and extremities.

c. Surgical excision. Must be used for management of soft-tissue/skeletal hypertrophy to address contour deformity.

D. Lymphatic malformations

- Slow-flow lesion
- Appearance

a. Anomalous lymphatic channels filled with lymphatic tissue which may be clustered into vesicles
b. Further classified as microcystic versus macrocystic

- Most common cause of macroglossia, macrochelia in children. Can also cause facial

asymmetry, distortion of surrounding tissue, soft- tissue/skeletal hypertrophy.

- Treatment options

a. Observation
   i. Intralesional bleeding can be treated with NSAIDS for pain control and rest.
   ii. Antibiotics indicated for cellulitis/other infections.

b. Sclerotherapy (mainstay treatment)
   i. Lesion is instilled with ethanol, doxycycline, sodium tetradecyl sulfate

ii. Can be used effectively when macrocysts are present (large volumes of lymphatic fluid without small loculations as seen on US or MRI). Microcystic disease, in which many small loculations are present, is not readily amenable to sclerotherapy.

c. Surgical resection

i. Direct excision can be effective but carries a high likelihood of significant complications.

ii. Suction-assisted lipectomy has been used to debulk lesions.

iii. Complications often include

- Cellulitis
- Hematoma
- Persistent drainage
- Recurrence of vesicular lesions
- Need for skin graft depending on extent of excision

E. Venous malformations

- Slow-flow lesion
- Appearance

a. Bluish, soft, compressible lesion which swells on dependent positioning; cluster of thin-walled veins with smooth muscle surrounding, many veins lack valves.

b. Changes in hormone levels can cause enlargement

- Must monitor for coagulopathy: Always perform coagulation profile studies as patients can develop DIC
- Treatment

a. Observation

b. Compression therapy

i. Useful for pain and edema

ii. Minimizes phlebothrombosis

c. Sclerotherapy: Most effective if performed on small cutaneous lesions using ethanol.

i. Side effects: Blistering, full thickness necrosis, neural deficits

ii. Make sure no important neural structures are nearby if using ethanol sclerotherapy.

d. Surgical excision: Performed most commonly after sclerotherapy to improve cosmetic appearance or remove mass tissue to optimize function.

F. Arteriovenous malformation

- High-flow lesion
- Appearance: Abnormal connections between arteries and veins

a. Arteries have thick, fibromuscular veins with prominent elastic lamina and stroma

b. Veins are "arterialized" and hyperplastic

- Clinical features
  a. Intracranial AVMs are more common than extracranial AVMs.
  b. *Always present at birth, but NOT always apparent. Puberty and trauma can stimulate enlargement
  c. Schobinger stages of development

    Stage 1: Quiescence—bluish discoloration and warmth of skin with AV shunting noted on Doppler examinations.

    Stage 2: Expansion—AVM begins to enlarge and demonstrates bruit, thrill, and pulsations.

    Stage 3: Destruction—begins to bleed, cause pain, and destroy surrounding tissue.

    Stage 4: Decompensation—persistent destruction to surrounding tissue with cardiac failure.
- Associated syndromes

  a. Bannayan–Zonana syndrome: AVM + microcephaly + lipomas

  b. Riley–Smith's syndrome: AVM + microcephaly + pseudopapilledema

  c. Osler–Weber–Rendu disease

    i. Multiple cutaneous telangiectasias with visceral AVMs (most commonly in lungs, liver, brain)
    ii. Often presents as frequent nosebleeds
- Treatment
  a. Observation: Used for small, clinically stable, asymptomatic lesions
  b. Embolization with surgical excision
    i. Used for Stage 3 to 4 lesions
    ii. Ligation/embolization of proximal feeding vessels must never be performed. Can cause recruitment of nearby vessels to exacerbate AVM.
    iii. Wide local excision is necessary to minimize recurrence rates.
    iv. Reconstruction with flaps is often necessary after excision.

HEMANGIOMA

- Vascular tumors that frequently appear within the first 4 weeks of life
- Affects females more commonly than males.
- Etiology: Benign proliferation of endothelial cells which is present after birth.
  - Most common tumor of infancy
  - More common in females (3:1)
  - Most common location: Head/neck region

- Stages of development—rapid postnatal growth, slow involution

1. Proliferating phase (0 to 12 months)
2. Involuting phase (12 months to 10 years)
3. Involuted phase (>10 years): Loose fibrofatty tissue replaces previous parenchymal tissue

*Approximately 50% of children experience complete involution by age 5.
*Approximately 70% experience complete involution by age 7.
*Minimal change expected after 12 years of age.

- Diagnosis
    - Mainly based on history and physical examination findings: Lesion presents after birth
    - and continues to enlarge in infancy.
    - Ultrasound (US).
    - MRI with contrast is useful for evaluation and demonstrate a well-defined vascular tumor

- Treatment options
    1. Initial observation
    2. Involution may be induced by steroid administration or propranolol treatment.
       *Cardiac monitoring is necessary for propranolol treatment
    3. Surgical excision is indicated for:
        i. persistent bleeding
        ii. when tumor location or growth impairs function or cosmesis (e.g., obstruction of globe, ear)
        iii. if the patient develops a platelet consumptive coagulopathy (Kassabach-Merritt syndrome).
    4. Laser ablation is also a possibility.