Neonatal Vascular Anomalies: *Evaluation*

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Relevant Financial Relationships

- I have no disclosures to make

Objectives

- Most vascular anomalies involve the skin and are noted at birth
- Review classification of vascular anomalies
- Discuss diagnostic imaging features of some of the most important vascular anomalies presenting in the prenatal and neonatal periods

Vascular Anomalies: ISSVA Classification

- Vascular tumors
  - characterized by endothelial hyperplasia
  - hemangiomas
  - less common tumors
- Vascular malformations
  - characterized by vascular dysmorphogenesis
  - normal endothelial cell turnover

Infantile Hemangioma

- Most common tumor of infancy
- Perinatal incidence of 1-2.6%
  - approximately 4% of all Caucasian infants affected in first year of life
  - lower incidence in dark-skinned infants
- Female-to-male ratio of 3:1 to 5:1
- Increased incidence in preterm infants weighing < 1000 gm (up to 30%)
- No clear genetic predisposition
- Risk factors
  - advanced maternal age, placental abnormalities, multiple gestations

Infantile Hemangioma

- 30-50% present at birth
- Cutaneous lesions permeate dermis
  - skin raised, bosselated, crimson in color
- Deeper lesions in lower dermis, subcutis or muscle often present as raised, bluish lesions with indistinct margins at 2-3 months of age or later

Infantile Hemangioma

- Located in head and neck (60%), trunk (25%), and extremities (15%)
- Increased risk of complications and need for treatment correlated with size
- About 80% solitary
- GLUT1-positive
- Infants with multifocal lesions more likely to have GI tract involvement
  - bleeding and anemia
- >5 lesions associated with increased risk of hepatic hemangioma
  - presentation 1-16 weeks postnatally with hepatomegaly, CHF, anemia or asymptomatic masses

Infantile Hemangioma: Clinical Course

- Proliferative phase
  - rapid growth during first 6-12 months of life
- Involuting phase
  - slow regression over 1-7 years
  - endothelial matrix replaced by loose fibrous or fibrofatty tissue
- Involuted phase
  - near-normal skin in about 50% of patients
  - telangiectasia, laxity, yellowish discoloration, scarring in remainder

Congenital Hemangioma

- Uncommon
- Evolves *in utero*
- Fully grown at birth
- Detected prenatally as early as 12th week of gestation
- Usually solitary
- GLUT1-negative
- Two types based on postnatal behavior
  - RICH (rapidly involuting congenital hemangioma)
  - NICH (non-involuting congenital hemangioma)

Hepatic Hemangioma

- Focal, multifocal, diffuse
- Focal lesions are the hepatic equivalent of cutaneous RICH
  - equal sex distribution, associated cutaneous infantile hemangiomas rare, GLUT-1 negative
  - involute over 10-23 months
- Multifocal and diffuse lesions are true infantile hemangiomas
  - female predominance, associated cutaneous infantile hemangiomas common, GLUT1-positive
  - rapid postnatal growth and slow involution over 1-5 years

Associated Malformative Anomalies

- PHACES syndrome
  - *P:* Posterior fossa and other structural brain anomalies
  - *H:* Hemangiomas of cervicofacial region
  - *A:* Arterial cerebrovascular anomalies
  - *C:* Cardiac defects, aortic coarctation and other aortic abnormalities
  - *E:* Eye anomalies
  - *S:* Sternal defects and/or Supraumbilical raphe
- Risk for stroke
  - MRI to assess brain and cerebral vasculature
- Ophthalmologic, endocrine and cardiac evaluation to rule out associated anomalies

Associated Malformative Anomalies

- Lumbosacral hemangiomas and occult spinal dysraphism
  - e.g. tethered cord, lipomeningocele
- Pelvic and perineal hemangiomas
  - urogenital and anorectal anomalies

Treatment

- Most hemangiomas small and regress without treatment
- Referral to specialty center in event of equivocal diagnosis, dangerous location, large size, rapidity of growth or potential for other complications
  - skin ulceration
  - CHF, hypothyroidism, abdominal compartment syndrome with hepatic hemangiomas

Kaposiform Hemangioendothelioma

- 60% present in neonatal period and 93% in infancy
- Unifocal
- Enlarging cutaneous lesion (75%), thrombocytopenia (56%), musculoskeletal pain or decreased function (23%)
- Affects trunk, shoulder, thigh or retroperitoneum
- Spectrum of clinical behavior and pathological findings
  - locally aggressive
  - slow-growing, benign ("Tufted angioma")

Vascular Malformations

- Localized or diffuse errors of embryonic development
- Affect about 1.2-1.5% of the population
- Most sporadic; some are inherited
- Affect any segment of the vascular tree
  - arterial, capillary, venous and lymphatic vessels
- Categorized according to predominant channel abnormality and flow characteristics
- Slow-flow anomalies
  - CMs, VMs, LMs
- Fast-flow anomalies
  - AVMs, AVFs
- Complex, combined vascular malformations
- No spontaneous regression

Venous Malformations

- Most common vascular anomaly
  - incidence of 1-2/10,000 births and 1% prevalence
  - occur throughout the body
    - head and neck (40%), extremities (40%), trunk (20%)
  - 95% sporadic
  - TIE2 mutation in some hereditary cutaneomucosal malformations
    - glomuvenous malformation most common
- Most are solitary
  - range from small, superficial and well-circumscribed lesions to large infiltrating lesions involving multiple soft –tissue planes
- Usually isolated lesion; some associated with syndromes

Lymphatic Malformations

- Localized or diffuse
- Dilated channels filled with proteinaceous fluid
- Generally not connected to normal lymphatic system
- Macrocystic (>1 cm), microcystic (< 1 cm) or combined
- Diagnosed prenatally, at birth or in early childhood
- Occur in head and neck (48%), trunk and extremities (42%), intrathoracic or intra-abdominal viscera (10%)
- Grow with child
- May enlarge rapidly after hemorrhage or infection

Arteriovenous Malformations

- Direct communication between dysplastic arteries and veins without intervening capillary bed
  - the shunt is the nidus of the AVM
  - high flow physiology
- Most are sporadic
- Heritable forms have been identified
- About 40% identified at birth
  - behavior unpredictable
  - usually dormant in infancy and childhood and enlarge during adolescence or following trauma or surgery
- Mass effect, soft tissue destruction, bone erosion

Complex Combined Malformations

- **Klippel-Trenauanay syndrome**
  - **may be due to somatic mutations in the *PIK3CA* gene**
  - **CLVM with soft-tissue and skeletal hypertrophy of limb(s) and/ or trunk**
- **CLOVES syndrome**
  - **somatic mutations in *PIK3CA* gene**
  - **congenital *l*ipomatous *o*vergrowth, *v*ascular malformations, epidermal nevi, *s*coliosis/skeletal and spinal anomalies**
- **Parkes Weber syndrome (PWS)**
  - **sporadic or inherited**
  - **large cutaneous CMs on extremity, multiple micro-AVFs and limb overgrowth**

Complex Combined Malformations

- **CM-AVM**
  - **Germline mutations in *RASA1* gene**
  - **CMs and high flow lesions (AVMs, AV fistulas, PWS)**

Summary

- **Reviewed classification of vascular anomalies**
- **Discussed some of most important lesions presenting in prenatal and neonatal periods**
- **Demonstrated critical role of imaging in the diagnosis of these abnormalities**