Neonatal Vascular Anomalies: 
*Evaluation*

Harriet J. Paltiel, MD
Boston Children’s Hospital
Harvard Medical School

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

Relevant Financial Relationships
- I have no disclosures to make
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
- 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
- Involuting 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, GLUT1 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 Supraventricular 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
• Glomovenous 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-Trenaunay 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 lipomatous overgrowth, vascular malformations, epidermal nevi, scoliosis/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