Current Classification And Nomenclature Of Vascular Malformations

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Classification of Vascular Anomalies

• Tumors
  • Hemangiomas
    • Infantile
    • Proliferating
    • Involuting
    • Involved
  • Congenital
    • Rich
    • NICH
    • Other tumors

• Vascular malformations
  • Simple
    • CM
    • VM
    • LM
    • AVM
  • Combined
    • CAVM
    • CLVM
    • CVM

Pathways involved in vascular anomalies

Pascal Brouillard, and Miikka Vikkula Hum. Mol. Genet. 2007;16:R140-R149

Infantile Hemangioma

• Rapid postnatal proliferation, slow involution
• GLUT1 positive
• Responds to angiogenesis inhibitors
Infantile Hemangioma

Multifocal (disseminated) Hemangioma

PHACES Association
- Post. Fossa anomalies
- Hemangioma, often plaque-like
- Arterial anomalies
- Cardiovascular anomalies
- Ear, Eye anomalies
- Sternal defects

Congenital Hemangioma
- fully grown at birth
- high flow
- rapid involution
Classification of Vascular Malformations

Venous Malformation

Incorrect terms:
- “cavernous hemangioma”
- “intramuscular hemangioma”

Venous Malformation; distends with Valsalva, dependency

Diffuse Venous Malformation

Soft, compressible

Focal VM

VM Morphology
Diffuse VM of the Limb
- All tissue planes are involved
- Consumption
- Coagulopathy
- Sclerotherapy for symptoms only
- Rapamycin

3 yo boy with severe consumption coagulopathy, pain
Glomovenous Malformations

Familial VM; TIE2 mutations; Blue Rubber Bleb Nevus Syndrome

3 week old with VM of Hand

Lymphatic Malformations

Macrocystic LM

Lymphatic Malformations
Microcystic Lymphatic Malformation

Diffuse Cystic LM

Types of Arteriovenous Malformations
Evolution of AVM; Schobinger Classification

Stage 1 quiescent
Stage 2 expansion
Stage 3 tissue ischemia

Diffuse AVM; progressive pain

Diffuse AVM; post amputation

Hereditary Hemorrhagic Telangiectasia
- Mutations in endoglin, ALK1 and SMAD4
- Nitric oxide synthetase defect -> progressive damage to the capillary bed

HHT

CM-AVM
- Autosomal dominant, 97% penetrance
- Atypical multifocal CM +/- AVM
- 30% have fast-flow lesion
  - Parkes Weber syndrome
  - Intracranial AVM / AVF
  - Facial AVM
PTEN Hamartoma Syndrome

- Cellular differentiation
- RAS system of tumor suppression
- Cowden’s syndrome
- Familial polyposis
- Bannayan Riley Ruvalcaba Syndrome
- ? Combined with HHT

Vascular Overgrowth Syndromes

- Sturge-Weber syndrome
- Klippel-Trenaunay syndrome
- Parkes Weber syndrome
- Proteus syndrome
- Maffucci’s syndrome
- CM-macrocephaly syndrome
PI3K, PTEN, mTOR pathways

Klippel-Trenaunay Syndrome
CLVM
• C = Capillary
• L = Lymphatic
• V = Venous Malformation
• With Overgrowth

KTS

Molecular Genetics of the PI3K–AKT–mTOR Pathway in Genodermatoses;