Selected Vascular Birthmarks

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Overview

- Segmental infantile hemangiomas
  - PHACE risk
  - LUMBAR risk
- Congenital hemangiomas
- Port-wine stains and Sturge-Weber risk
- Vascular stains and overgrowth
Using a systematic (checklist) approach

- **Which** to worry about
- **What** to do
- **When** to do it
- **Trying to avoid** overdoing
  - MRI – need for GA
  - Costs – both financial and emotional
PHACE risk: What to do

- ~30% chance of PHACE hemangiomas > 5 cm diameter especially segments 1, 3 and 4

✓ Complete PHACE w/u = MRI/MRA head and neck, cardiac echo, eye exam

Key Article re: PHACE

Updated consensus criteria

- All infants with large segmental IH located on either the face or scalp
- PHACE should be considered with 1 major criterion of PHACE and a large segmental hemangioma of the neck, upper trunk, or trunk and proximal upper extremity
- 2 major criteria of PHACE (eg, supraumbilical raphe and coarctation of the aorta) but lacking cutaneous IH
Beyond the highest risk group:

- S2 and parotid IH are lower risk
  - Partial work-up e.g. eye, echo, less sure about MRI/A

- Other risk settings for PHACE
  - Large periorbital IH even if not extending to S1
  - Large torso plus arm IH
  - Segmental scalp

Key Point

- Severe coarctation of aorta is an absolute contraindication to propranolol therapy
- For patients at higher risk for PHACE, echocardiogram is strongly recommended
  - Need to get cardiology consultation
  - Can be done without sedation
Cerebrovascular Risk

- Concerns re: propanolol in at-risk patients of possibly provoking CNS ischemia/stroke

- The reality:
  - Risk is probably very low
  - Most patients with PHACE need propranolol therapy to manage their hemangiomas

- Need to risk-stratify degree of CNS arteriopathy

Siegel et al. Stroke. 2012;43:1672-4
<table>
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<tr>
<th>Risk Category</th>
<th>Cerebrovascular Anomalies</th>
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| **High**     | • Multiple vessels with severe narrowing or non-visualization without adequate collateral circulation and Moyamoya disease and Cardiac or Aortic arch anomalies  
• Severe narrowing/stenosis\(^b\) or non-visualization of 1 major vessel\(^c\) without adequate collateral circulation and Moyamoya disease and Cardiac or Aortic arch anomalies  
• Severe narrowing/stenosis\(^b\) or non-visualization of 1 major vessel\(^c\) without adequate collateral circulation and Moyamoya disease  
• Severe narrowing/stenosis\(^b\) or non-visualization of 1 major vessel\(^c\) without adequate collateral circulation and Cardiac or Aortic arch anomalies  
• Severe narrowing/stenosis\(^b\) or non-visualization of 1 major vessel\(^c\) without adequate collateral circulation |
| **Intermediate** | • Severe narrowing/stenosis\(^b\) of major vessels\(^c\) *with* adequate collateral circulation  
• Mild narrowing/stenosis\(^d\) of major vessels\(^c\) *with* adequate collateral circulation  
• Hypoplasia, dysplasia, aberrant origin or course of major vessels\(^c, e\)  
• Persistent embryonic arteries |
| **Low**       | • No arterial anomalies |
My “mental check-list” if patients have PHACE or at-risk for PHACE

- Neuro status (ideally with neurologist) especially
  - Motor development
  - Language development
- Hearing test (not just newborn screen)
- Dental development
- Somatic growth
- Evidence of headaches
- Late hemangioma growth
LUMBAR syndrome

- MRI (not ultrasound) with/without contrast needed for adequate exam of spine
- Renal u/s or other evaluations not standardized
  - Depends on clinical setting

Rapidly-involuting Congenital Hemangioma (RICH)

- Vascular tumor present at time of delivery
- Rapid improvement in first few months of life
- Usually a clinical diagnosis
- Morphology can vary
RICH: Checklist

- Is this the correct diagnosis?

- Coagulopathy Risk
  - Check CBC with platelets and D-Dimers

- Ulceration or impending ulceration:
  - Be aware of potential life-threatening bleeding

- Even rarer: associated high-output cardiac state

- Watch over time for expected improvement
  Resolution may be partial (i.e. PICH)
From: Life-Threatening Hemorrhaging in Neonatal Ulcerated Congenital HemangiomaTwo Case Reports

Vascular Stains

- Previous all lumped under “port-wine stain” (or “capillary malformation”) but now can see distinct differences and risks

- Key article:

Nevus Simplex Complex

- Mid-line and characteristic sites (vs PWS)
- Spontaneous resolution...*usually*
- Most are *not* associated with extra-cutaneous dz
  - Certain exceptions

Extracutaneous associations

- Nevus-simplex like stain overlying developmental anomaly (e.g. lump, lipoma, hypertrichosis, aplasia cutis)
- Megancephaly-Capillary Malformation (M-CM)
- Beckwith Weidemann syn
- Roberts SC (Pseudo-thalidomide syndrome)
- Nova syndrome
Port-wine stains of SWS patients

- 11/66 (16%) patients with upper facial PWS had SWS
- Forehead confers risk – most not classic V1


Worried about SWS: What to do?

- Eye exam always if eyelid involvement
  – upper or lower

- Imaging – not so clear - since may not change management

- Family awareness re: focal motor seizures

- If very extensive/high-risk (e.g. hemifacial or bilateral) consider neuro referral and MR-imaging
Exam Essentials

- Head circumference (clue is "big forehead")
- Presence of nevus simplex/scalp or facial dysmorphism
- Body asymmetry/overgrowth
- Digits: overgrowth, syndactyly, splaying of digits (sandal gap)
Diffuse Capillary Malformation with Overgrowth (DCMO)

- Have proportionate rather than progressive overgrowth
- ~ one-third have varicosities
- Associated with GNAQ/GNA11 somatic mutations

Widespread blotchy Stains with Overgrowth

PLUS

✓ If large head and/or digital anomalies: Multidisiciplinary evaluation
  ✓ Genetics consult
  ✓ Consider somatic gene testing
  ✓ Neuro evaluation
  ✓ Consider brain MRI
Widespread stains minus worrisome features

✓ If lower extremity involvement: Serial leg measurements until growth completed
  ✓ If > 1-2 cm refer to orthopedics

✓ If no worrisome features:
  ✓ Initial f/u 3-6 months
  ✓ Thereafter annually
Geographic stains

- All borders more sharply demarcated
- Frequent presence or development of blebs
- Less blanchable than blotchy PWS
- Suggest lymphatic disease
- Sign of PIK3CA overgrowth syn (PROS)
Geographic stains Checklist

- Watch over time for overgrowth and blebs
- Multidisciplinary care strongly advised!
- MRI with and without contrast
- Lymphatic complications
  - Warn about blebs
  - Risk of infection
- Wilms tumor surveillance? (for CLOVE)
- Coagulopathy risk
Multifocal Stains: Approach

- Always ask about family history
- Check capillary refill in stains
- Most (~90%) CM-AVM due to mutations in RASA-1
  - Autosomal dominant
  - 10% risk of spinal or brain AVM
- Genetic referral/testing
- Imaging of brain and spine…when and how often unclear

Revençu N et al Hum Mutat. 2013;34:1632-41