Agenda

• Index Patient Introduction
• Disease Classification
• Disease Descriptions
• Imaging Workup
• Treatment Options
• Interventional Management
Index Patient: Baby B.G.

• History
  • 2 mo M with left neck mass first seen prenatally at 37wk U/S. Pt born via c/s at 39wk, otherwise unremarkable birth. Lesion has not changed size since birth. No problems with growth, airway, feeding.

• Physical Exam
  • AVSS, boggy, 5x6cm left neck mass, no overlying rash, warmth, or bruit.

• Brief Differential Diagnosis
  • Vascular anomaly – most likely
  • Infection - smaller, transient, usually after birth
  • Solid tumor - (benign/malignant) – usually firm, midline
  • Branchial cleft cyst - later age, firm, smaller
  • Thyroglossal duct cyst - later age, central location
ISSVA Classification

- ISSVA - Int’l. Society for the Study of Vascular Anomalies
  - Drs. Mulliken & Glowacki - Children’s Hospital Boston
- Biologic Classification – differing course & treatments

  **Tumor vs Malformation**

  - “oma” = proliferation
  - ↑ EC turnover / hyperplasia, thick BM
  - ↑ Surface markers - VEGF, bFGF, (PCNA)
  - Usually infancy/childhood
  - Naturally involuting
  - >3:1 female:male

  - abnormal morphogenesis
  - Normal EC, BM, pathology
  - Minimal surface marker expression
  - Present at birth
  - Naturally persistent
  - 1:1 female:male
Classes of Congenital Vascular Anomalies

- **Tumors**
  - **Hemangiomas**
    - Infantile
    - Congenital
  - **Tufted Hemangioma**
  - **Hemangioendothelioma**
  - **Acquired dermatologic**
  - **Other syndromes**

- **Malformations**
  - **Capillary**
    - Dermatologic
    - Superficial laser tx
  - **Lymphatic**
    - Microcystic
    - Macrocystic
  - **Venous**
  - **Arterial / Arteriovenous**
  - **Combined Forms / Syndromes**
Hemangioma

- Benign endothelial cell tumor
  - Tightly packed mass of vascular channels’
  - 2 main types
- 1. **Infantile Hemangioma**
  - Usually has overlying patch of redness (superficial)
  - Most common tumor of infancy/childhood
    - 4-10% prevalence in Caucasian infants
    - 3-5:1 females:males
  - Appears weeks/months after birth
  - Natural course - 3 stages
    - 1. Proliferating - first year
    - 2. Involuting - few years
    - 3. Involuted - most resolved by age 10

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Hemangioma (cont.)

2. **Congenital Hemangioma**
   - Blue/gray hue w/ pale halo (skin)
   - Rare (compared to infantile)
   - Present at birth
   - 2 types
     - 1. Non-Involuting (NICH) - persistent
     - 2. Rapidly Involuting (RICH) - resolved by 1-2 yrs

Complications
- Ulceration, bleeding, infection, obstruction/displacement of organs, high-output cardiac failure due to high flow/shunting

There are NO new-onset adult hemangiomas
Lymphatic Malformation (LM)

- Collection of lymph-filled channels/cysts
- Present at birth (5-6 wks G.A.)
- ↑ Swelling w/ infections
- Soft w/ no overlying rash
- Most common:
  - 1. head/neck
  - 2. extremities/axilla
  - 3. trunk
- 2 Types
  - 1. Microcystic: multiple small vesicles
  - 2. Macrocystic: few large septated cysts
- Complications: infection, bleeding, obstruction/displacement of organs, overgrowth of involved tissue
- A.K.A. - “cystic hygroma”, “lymphangioma”

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http://www.childrenshospital.org/az/Site1256/mainpageS1256P0.html
Venous Malformation (VM)

- Thin-walled, dilated veins
  - Inadequate smooth muscle layer
- Present at birth
  - Often unseen until symptomatic in childhood
- Soft w/ bluish skin hue
- Waxing/waning size and symptoms
  - VM growth proportional to child’s growth
  - Possible association with trauma, hormones
- Complications
  - Thrombosis, bleeding
- A.K.A. - “cavernous hemangioma”

Children’s Hospital Boston
http://www.childrenshospital.org/az/Site1830/mainpageS1830P0.html

Companion Patients #2 and #3
Arterio-Venous Malformation (AVM)

- High-flow arterio-venous communication - absence of developed capillary bed
- Present at birth
- Reddish vascular hue (skin), often warm
- Complications
  - Bleeding, compression / displacement of organs, high-output cardiac failure
- Seen in hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu)

http://www.childrenshospital.org/az/Site593/mainpageS593P0.html
Baby B.G. - Focused DDx

- **Review**: soft left neck mass since birth, no change in size, no warmth/redness

- **Narrowed Differential Diagnosis?**
  - Vascular anomaly
    - Hemangioma?
      - Infantile? - No - present since birth
      - Congenital? – Possible – too soon to distinguish NICH vs RICH
    - Lymphatic? – Possible
    - Venous? - Less likely but possible - no growth but only 2 months old, no bluish hue but not always present
    - Arterial / AV? - Less likely - no warmth/redness
With a focused differential diagnosis based on history and physical, we proceed to radiologic imaging to further characterize our patient’s vascular malformation.
Imaging Options for Vascular Malformations

- **Ultrasound**
  - Assess flow pattern
- **MRI**
  - Critical, often definitive
- **Radiographs**
  - Limited benefit - bony structures, calcification
  - Quick and Cheap
- **Angiography**
Imaging Workup Decision Tree

Ultrasound

MRI

High flow

Mass-like

Hemangioma

No mass

AVM

Low flow

Diffuse enhancement with contrast

Venous

No/rim enhancement with contrast

Lymphatic
Ultrasound Reference Images

- **High Flow Lesions**
  - Hemangioma
  - Arterio-venous Malformation

- **Low Flow Lesions**
  - Lymphatic Malformation
  - Venous Malformation
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MRI Images - High Flow Lesions

- **Hemangioma**
  - Protruding mass (*)

- **Arterio-Venous Malformation**
  - No mass
  - Flow voids – high-speed flow

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MRI Images - Low Flow Lesions

- **Venous Malformation**
  - Diffuse enhancement w/ contrast

- **Lymphatic Malformation**
  - Septal (Left) / Rim (Rt) enhancement

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Radiographic / (CT) Findings

- Generally of limited use
- Phleboliths seen w/ X-ray
  - Calcifications
    - Post venous thrombus
  - Suggest Venous malformations

Left upper extremity

Angiography

- Performed to characterize AVM architecture
- Encouraged prior to any injected therapy
- No longer necessary for diagnosis of venous malformation
Baby B.G.’s Radiologic Studies and Diagnosis

Macrocystic Lymphatic Malformation

- No phlebolith
- Low flow
- Large cysts
- T1 hypointense
- Final Diagnosis?

Axial MRI L Neck, T1 (contrast study not performed at outside referring hospital)

Axial MRI L Neck, T2

Ultrasound L Neck

Ultrasound w/ Doppler
Further Workup Options

- Biopsy
  - Pathology / Microbiology
- Aspirate of lesion
  - Blood vs Lymph
  - Pathology / Microbiology
- Molecular Markers
  - Of lesion sample
  - Of patient’s serum/urine
Treatment Options

- Observation
- Dermatologic
  - Laser therapy – capillary malformation
- Pharmacologic
  - Hemangioma – steroids, IFN-α, vincristine
- Surgical (excision)
- Interventional Radiology (IR): minimally invasive
  - **Sclerotherapy** - LM & VM (low-flow lesions)
  - Embolization - AVM
Sclerotherapy Overview

- Primary IR treatment for VM/LM
- Intralesional injection of irritant/sclerosant
  - U/S & fluoroscopically guided
  - Induces fibrosis, contraction over 4-8 weeks
- Sclerosants
  - Doxycycline: sufficient for LM
  - Bleomycin: experimental for microcystic LM
    - Theoretical concern for systemic effects – pulmonary fibrosis
  - Sodium Tetradecyl Sulfate (STS): detergent for VM/LM
  - OK-432: experimental, lyophilized S. pyogenes cells
  - EtOH: avoided in children
Sclerotherapy Setup

Dr. Konez, http://www.birthmarks.us/sclerotherapy.htm
U/S Pre & Post VM Sclerotherapy

U/S Normal Muscle
Fascicular horizontal lines noted

U/S Pre STS
Venous channels circled, fibrotic (grainy echogenicity) muscle surrounding

4-6wks Post Sclero
Reduced channel size, sclerotic/fibrotic muscle surrounding

PACS, CHB, courtesy Dr. C. Johnson
Baby B.G. Sclerotherapy: Left Neck Ultrasound

Lymphatic Macrocyt (Fluid = black)

Injection Doxycycline after fluid aspiration

U/S guided needle insertion

Post injection

PACS, CHB, courtesy Dr. Padua
Baby B.G. Sclerotherapy: Fluoroscopy

Fluoroscopy – Doxycycline injection of cyst

Post injection with contrast/sclerosant filled cyst
Representative images of neck lymphatic malformation pre and 2 years post sclerotherapy with doxycycline
Pre & Post Sclerotherapy for VM

Companion Patient #5

Children’s Hospital Boston
http://www.childrenshospital.org/az/Site1830/mainpageS1830P0.html
Summary

- **Classification**: Tumor vs Malformation
- **Imaging Workup**
  - 1. Ultrasound - High vs Low Flow
  - 2. MRI - T2/T1, contrast enhancing (blood), flow voids (high flow)

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- **Treatment**: Pharmacologic, Surgical, Interventional
  - Hemangioma - Steroids
  - Lymphatic Malformation - Surgery / Sclerotherapy (Doxycycline)
  - Venous Malformation - Surgery / Sclerotherapy (STS)
  - Arterial - Embolization
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References

Thank You

Questions?
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