Cases from the Vascular Birthmark Clinic

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Disclosures
• None of the three speakers have any disclosures.
• Some of our photos are of our VBMC patients. All of their families have given informed written consent for photographs for teaching.
• We will discuss an off label use for topical timolol.

Educational Objectives
• Present new classification of vascular anomalies
• Describe the appropriate use of VMBC service for MD advice/referral and how to formulate a plan for who needs referral
• Learn how to recognize some of the syndromes found in the classification of vascular anomalies and which need referral

Overview of Vascular Birthmark Types

Case Reports
• Hemangioma
• Port Wine Stain
• Venous Malformation
• Lymphatic Malformation
• Arteriovenous Malformation
Hemangioma Case- L.H.
• Born at 38 weeks in Bakersfield
• 10 days-noticed “lipstick” on left lower lip. Grew rapidly
• 2 mo-bleeding, ulceration. PCP contacts us, rec clobetasol and aquaphor until we can see her the following week
• 2 mo-VBMC, PDL, endoscopy by HNS, no lesions seen
• 3 1/2 mo-admitted to LAMC for Propranolol-improvement
• 12 mo-weaned off propranolol-residual hemangioma

Delayed Surgical Consultation

Before and After

EH- Infant on Steroids

After Surgical Debulking

Case Reports
• Hemangioma
• Port Wine Stain
• Venous Malformation
• Lymphatic Malformation
• Arteriovenous Malformation
Port Wine Stain Clinical Case  
**LC, 16year old Female**

**Problem list:**  
- Sturge Weber Syndrome  
- Asthma  
- Scoliosis  
- Musculoskeletal anomaly  
- Right eye glaucoma associated with vascular disorder  
- Hx of eye removal  
- Complex partial epilepsy on carbetrol, keppra  
- Developmental delay, global  
- GH deficiency  
- Short Stature  
- Klippel Trenaunay Syndrome  
- Skeletal malocclusion  
- Lordosis of spine  
- Right infantile and juvenile cataract  
- Blind left eye, low vision right eye  
- Legal blindness  
- Profound intellectual disability

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Port Wine Stain case

**Port Wine Stain case**

**Port Wine Stain case**

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Port Wine Stain Clinical Case  
**VM, 40year old Female**

**Problem list:**  
- Sturge Weber Syndrome  
- Port wine stain at upper left lip, cheek, upper eyelid, hypertrophy of left cheek, hypertrophy of left lip  
- Hemihypertrophy of entire left body (same side as facial port wine stain/hypertrophy)  
- Larger breast left side (“D” cup) compared to right breast (“A” cup), s/p breast augmentation  
- Right breast and reduction of left breast  
- Left eye hypertension  
- New 4/11/14: jerky at right arm episode, short but repetitive  
- Neurology advised that she has probably has done this since she was younger (she called them “zone outs”) but thought it was normal  
- Started on Keppra 2014  
- She was advised that she had Sturge Weber syndrome in 2014
Port Wine Stain case

• Present at birth
• Occur equally in both sexes
• Prevalence rate is 0.3-0.5%
• Lesions tend to grow with the patient
• No tendency towards regression
• Usually located on the face, V2 dermatome is the most common site

Port Wine Stain case

• Initially appear as light-pink macules
• Can darken over time as a result of progressive vessel ectasia
• Overlying or bony hypertrophy may occur
• PDL is the preferred treatment
Port Wine Stain

Patients treated earlier in the natural history have increased efficacy and decreased likelihood of recurrence.

Lesions on the head and neck location and small lesions (<20cm) respond better to laser treatment.

Pulsed Dye Laser (PDL)

- 595nm wavelength, chromophore target is hemoglobin
- Selective photothermolysis, 1.5mm depth, cryogen cool spray

Pulsed Dye Laser (PDL)

• Lesions tend to fade by 80% after minimum of 8 to 10 treatments
• PWS on the face or neck tend to respond more quickly than those on the lower extremities
• Expect transient edema and purpura, can last 2 to 7 days.

Sturge Weber Syndrome
Sturge Weber Syndrome

- Aka: Encephalotrigeminal angiomatosis
- Neurocutaneous disorder
- Triad: cutaneous (port wine stain V1/upper eyelid 96%), neurologic (seizures 75-90%) and ocular (glaucoma 30-71%)
- Abnormal capillary venous vessels in the leptomeninges of the brain and choroid, glaucoma, seizures, stroke and intellectual disability
- Incidence in US: 1 in 50,000

Sturge Weber Syndrome

Triad:
- Cutaneous (port wine stain V1/upper eyelid 96%)
- Neurologic (seizures 75-90%)
- Ocular (glaucoma 30-71%)

Sturge Weber Syndrome

Pathophysiology
- Residual embryonal blood vessels and their secondary effects on surrounding brain tissue.
- The vascular plexus that normally regresses in the 9th week gestation fails to regress results in residual vascular tissue forms angiomata of the leptomeninges, face and ipsilateral eye.
- Sporadic, somatic activating mutation in GNAQ

Sturge Weber Syndrome

Classification:
- Type I: Facial and leptomeningeal angiomas, may have glaucoma
- Type II: Facial angioma alone (no CNS), may have glaucoma
- Type III: Isolated LA, usually no glaucoma

Sturge Weber Syndrome

Physical signs of SWS:
- Portwine stain
- Macrocephaly
- Ocular manifestations
- Soft tissue hypertrophy
- Hemiparesis
- Visual loss
- Hemianopsia
Sturge Weber Syndrome

- Physical signs of SWS:

Skull x-ray /CT:
- "tram tracks"

CT: gyriform pattern of subcortical calcifications

MRI with and without contrast:
- Atrophy of the cerebral cortex, abnormal myelination,
Sturge Weber Syndrome

**Treatment**
- Symptomatic
- Laser to lighten or remove the birthmark
- Anticonvulsant medications for seizures
- Surgery for glaucoma
- Physical therapy for infants and children with muscle weakness
- Educational programs for mental retardation or developmental delays.
- Yearly monitoring for glaucoma

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**Case Reports**

- Hemangioma
- Port Wine Stain
- **Venous Malformation**
- Lymphatic Malformation
- Arteriovenous Malformation

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**Venous Malformation Case**

- Child presented to the vascular birthmark clinic at age 6.
- Born with purple spots over his left knee/thigh.
- Was told at a children’s hospital that they would disappear over time.
Venous Malformation

- Over time, the lesions became more prominent with pain, swelling and warmth
- Symptoms worsened with 15 minutes of activity, standing for long periods of time and during hot weather
- Symptoms helped with rest, elevation and Ibuprofen

Venous Malformation: MRI

- Prominent veins in the affected area
- Does not follow anatomic planes
- Ill-defined borders
- Can have prominent draining veins
- No arterial feeders
- Can sometimes see phleboliths

Venous Malformation: Venogram

Venogram:
- Prominent, abnormal veins

Venous Malformation Progression

- Over time, the lesions became more prominent with pain, swelling and warmth
- Symptoms worsened with 15 minutes of activity, standing for long periods of time and during hot weather
- Symptoms helped with rest, elevation and Ibuprofen

Venous Malformation: Sclerotherapy

Sclerotherapy:
- Inject a sclerosant into the abnormal veins to cause fibrosis.
- Prevents dilatation of the vein.
- Can perform multiple times.

Treatment options:
- NSAIDS
- Compression stockings
- Sclerotherapy
- Surgical resection
Venous Malformation

- Patient now 12 years old
- Has been treated 3 times in 6 years
- Has pain and/or swelling a few days a week
- Wears a compression stocking as needed, ibuprofen as needed
- Will consider repeat sclerotherapy when patient and mother decide it is severe enough to pursue

Case Reports

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Lymphatic Malformation

- 19 month old girl, previously healthy, presented with left chest/axillary swelling after falling on a toy 2 months ago.
- No pain, erythema, warmth, or fever. Growth of lesion now affects arm positioning and causes irritation.

Lymphatic Malformation

- Exam:
  - Soft, partially compressible large bulge in the left axilla
  - Not pulsatile
  - No prominent veins

Lymphatic Malformation- MRI

- Large cystic space in the subcutaneous tissues of the left chest wall
- Features typical of water
- Macrocytic, unilocular
- Options:
  - Surgical resection
  - Sclerotherapy
  - Sirolimus

Lymphatic Malformation

- Patient’s parents chose to undergo sclerotherapy.
- Lesion accessed with a needle
- For large lesions, a drain is left in place
- Cavity injected with sclerosant
- May require multiple visits
Lymphatic Malformation

- Cavity resolved after 3 treatments in 3 weeks

Lymphatic Malformation

- Complete resolution of bulge

Case Reports

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AVM

- 11 year old with a bump on her forehead since 1.5 years old after a fall.
- Pulsatile sensation
- Growing
- Avid dancer. More prominent after activity
- 4 years ago in Oregon, diagnosed with a vascular malformation

AVM

- Exam:
  - Left medial forehead 1.5 x 2 cm mass
  - Partially reducible
  - Pulsatile, with palpable thrill

CTA

- Focal prominent vasculature
- Possible feeding artery/draining veins
AVM- Treatment Options

• Embolization
• With surgical resection afterwards
• Conservative therapy generally not recommended

AVM

• Angiography
  • Feeding arteries from branches of both the internal and external carotid arteries

AVM

• Embolization
  • Typically done with glue or Onyx

AVM

• No residual flow to the lesion

AVM- Surgical Excision

• Following embolization, the patient’s symptoms resolved, but she still had a bump.
• Patient and her family opted for surgical excision for cosmesis
• Failure to completely thrombose the AVM will result in its eventual recurrence, which could be complex and difficult to treat.
Summary

- **Venous/lymphatic malformations**
  - Usually not curable
  - Treatment for symptomatic control
  - Treatment options
    - NSAIDS
    - Compression stockings
    - Sclerotherapy
    - Surgical resection

- **Arteriovenous malformations**
  - Will worsen over time if left untreated
  - Treatment:
    - Embolization
    +/- surgical resection

Vascular malformation websites

- [www.issva.org/](http://www.issva.org/)
- [www.cincinnatichildrens.org/health/v/maformation/](http://www.cincinnatichildrens.org/health/v/maformation/)
- [www.childrenshospital.org/health-topics/conditions/vascular-malformations-tumors-and-hemangiomas](http://www.childrenshospital.org/health-topics/conditions/vascular-malformations-tumors-and-hemangiomas)
- [http://www.chop.edu/conditions-diseases/vascular-malformations](http://www.chop.edu/conditions-diseases/vascular-malformations)
- [http://www.novanews.org/information/vascular-malformations](http://www.novanews.org/information/vascular-malformations)

References:


Questions?