Congenital vascular syndromes: diagnostic role of a multidisciplinary clinic

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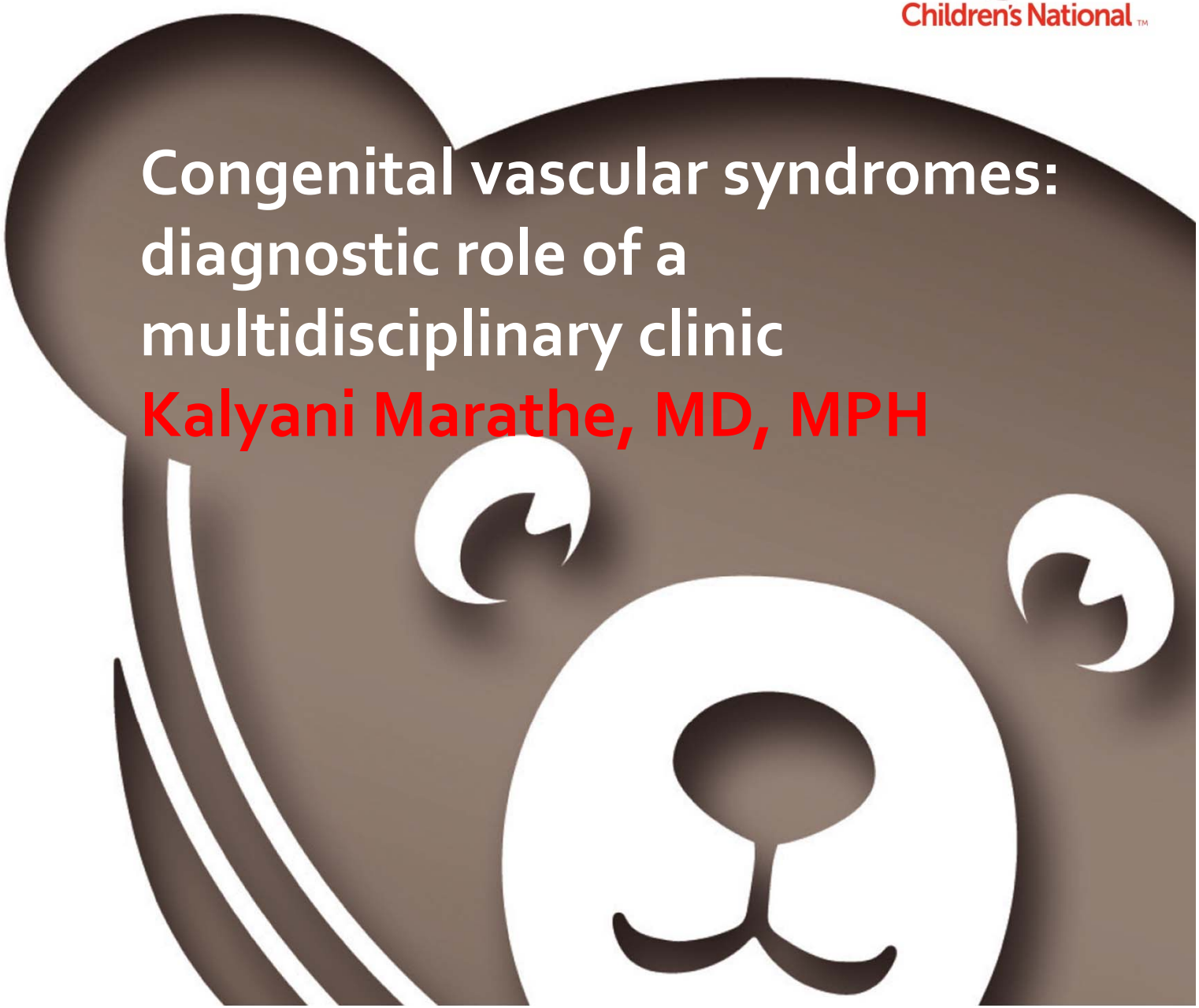

I have no conflicts of interest or relevant financial relationships to discuss.

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Objectives

At the end of this talk, audience members should be able to:

- Identify PHACE and Sturge-Weber syndrome based on their clinical characteristics
- Recognize associated features
- Initiate an appropriate workup

Case 1

- A 2-week-old girl presents to clinic with the following lesion:
- What are the most important components of a workup?

Photo, Bolognia 3rd ed.c

Case 2

- A 2-week-old girl presents to clinic with the following lesion:
- What are the most important components of a workup?

Photo courtesy of A. Yasmine Kirkorian MD

Case 3

- A 2-week-old girl presents to clinic with the following lesion:
- What are the most important components of a workup?

Photo courtesy of A. Yasmine Kirkorian MD

ISSVA

- International Society for the Study of Vascular Anomalies
- Organizes biennial workshops
- Classification scheme
  - Vascular malformations
    - Capillary malformations
    - Venous malformations
    - Lymphatic malformations
    - Arteriovenous malformations
  - Hemangiomas

Appendix 2-a
causal genes of vascular anomalies

Capillary malformations (CM)	
Cutaneous and/or mucosal CM (aka “port-wine” stain)	GNAQ
CM with bone and/or soft tissue hyperplasia	
CM with CNS and/or ocular anomalies (Sturge-Weber syndrome)	GNAQ
CM of CM-AVM	RASA1
Telangiectasia	
Hereditary hemorrhagic telangiectasia (HHT)	
HHT1	ENG
HHT2	ACVRL1
HHT3	
JPHT (juvenile polyposis hemorrhagic telangiectasia)	SMAD4
Others	
Cutis marmorata telangiectatica congenita (CMTC)	
Nevus simplex / Salmon patch	
Others	

Sturge Weber Syndrome

- Syndromic capillary malformation with CNS and/or ocular abnormalities
- Unilateral facial CM; usually involves forehead and upper eyelid, may be bilateral

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- If both upper and mid face involved, ocular involvement more likely
  - Congenital glaucoma
  - Choroidal vascular malformation

Sturge Weber Syndrome

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- Unilateral facial CM; usually involves forehead and upper eyelid, may be bilateral
- If both upper and mid face involved, ocular involvement more likely
  - Congenital glaucoma
  - Choroidal vascular malformation
- CNS involvement: due to leptomeningeal vascular malformations
  - Seizures (typically develop in first year of life)
  - Less commonly, hemiparesis/hemiplegia, developmental delays, emotional/behavioral

Workup for Sturge-Weber Syndrome

- Pediatric Ophthalmology evaluation at birth
  - Follow up regularly, as glaucoma may not become evident until childhood
- Pediatric Neurology referral
  - MRI if symptomatic

ISSVA Classification

Appendix 3

infantile hemangioma

Pattern	Different types
- focal - multifocal - segmental - indeterminate	- superficial - deep - mixed (superficial + deep) - reticular / abortive / minimal growth - others

Association with other lesions	
PHACE association / syndrome	Posterior fossa malformations, Hemangioma, Arterial anomalies, Cardiovascular anomalies, Eye anomalies, sternal clefting and/or supraumbilical raphe
LUMBAR (SACRAL, PELVIS) association / syndrome	Lower body hemangioma, Urogenital anomalies, Ulceration, Myelopathy, Bony deformities, Anorectal malformations, Arterial anomalies, and Renal anomalies

Case 2

- A 2-week-old girl presents to clinic with the following lesion:
- What are the most important components of a workup?

PHACE(S) Syndrome

- **P**osterior fossa malformations
- **H**emangioma (segmental)
- **A**rterial anomalies
- **C**ardiac anomalies/**C**oarctation of aorta
- **E**ye anomalies
- (**S**upraumbilical raphe/**S**ternal clefting)

Arch Dermatol. 1996 Mar;132(3):307-11.

PHACE syndrome. The association of posterior fossa brain malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, and eye abnormalities.

Frieden IJ[1], Reese V, Cohen D.

PHACE(S) Syndrome

- **P**osterior fossa malformations
  - Dandy-Walker malformation
- **H**emangioma (segmental)
- **A**rterial anomalies
  - Head and neck: stenosis, tortuosity, aberrance
- **C**ardiac anomalies/**C**oarctation of aorta
  - PDA, ASD, VSD, Tetralogy of Fallot
- **E**ye anomalies
  - Horner syndrome, retinal vascular anomalies
- (**S**upraumbilical raphe/**S**ternal clefting)
  - Ventral midline developmental defects

Workup for PHACE

- Neuroimaging
  - MRI/MRA with TRICKS protocol
- Echocardiogram
- Ophthalmologic evaluation

Appendix 2-e
causal genes of vascular anomalies

Vascular malformations associated with other anomalies	
Klippel-Trenaunay syndrome	
Parkes Weber syndrome	RASA1
Servelle-Martorell syndrome	
Sturge-Weber syndrome	GNAQ
Limb CM + congenital non-progressive limb overgrowth	
Maffucci syndrome	
Macrocephaly - CM (M-CM or MCAP)	PIK3CA
Microcephaly - CM (MICCAP)	STAMBP
CLOVES syndrome	PIK3CA
Proteus syndrome	AKT1
Bannayan-Riley-Ruvalcaba syndrome	PTEN

Photo courtesy of A. Yasmine Kirkorian, MD

LUMBAR syndrome

- **L**ower body congenital infantile hemangiomas and other skin defects
- **U**rogenital anomalies and ulceration
- **M**yelopathy
- **B**ony deformities
- **A**norectal malformations/Arterial anomalies
- **R**ectal anomalies

Workup for LUMBAR

- At < 3 months
  - Spinal ultrasound
  - Ultrasound with doppler- abdomen and pelvis
- 3-6 months
  - If midline lesion or abnormal ultrasound, MRI of spine
  - If abnormal pelvic/renal ultrasound, urologic evaluation
  - Monitor for limb length discrepancy with Orthopedics

Thank you!!!!!

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