

# PHACE Syndrome Family Conference

Maria C. Garzon, M.D.

Professor of Dermatology and Pediatrics at CUMC

Columbia University, NY



**New York-Presbyterian  
Morgan Stanley Children's Hospital**



**Columbia University, NY**

## **DISCLOSURE OF RELATIONSHIPS WITH INDUSTRY , CONFLICTS OF INTEREST**

Maria Garzon, M.D.

### DISCLOSURES

I do not have any relevant relationships with industry.

# phace syndrome™ community



# Consensus Derived Diagnoses and Care Recommendations

- Diagnostic criteria established in 2009
- New information and need for an update
- Interdisciplinary research meeting

# Medical Progress in PHACE syndrome: Consensus Derived Diagnoses and Care Recommendations

- PHACE syndrome workshop held in Milwaukee Wisconsin June 2014
  - 28 physicians from 14 different institutions
  - 9 different types of sub-specialists
  - Parents and advocates
- Clinical experience, review of the existing medical literature on PHACE
- Specialty focused working groups
  - Content presented at the meeting for discussion
  - Expert participants were asked to reach consensus and develop recommendations regarding organ specific screening protocols and risk adjusted surveillance strategies.
- Recommendations were then presented to the larger multidisciplinary group, refined and edited until consensus was reached.
- Protocols were collated re-reviewed by the expert participants
  - Conference calls and electronically

## Medical Progress in PHACE syndrome: Consensus Derived Diagnoses and Care Recommendations

Maria C. Garzon\*, MD; Departments of Dermatology and Pediatrics, Columbia University  
Leon G. Epstein, MD; Departments of Pediatrics and Neurology, Northwestern University Feinberg School of Medicine  
Geoffrey L. Heyer\*, MD; Departments of Pediatrics and Neurology, Nationwide Children's Hospital and The Ohio State University  
Peter C. Frommelt, MD; Department of Pediatrics, Medical College of Wisconsin  
Darren B. Orbach\*, MD; Division of Neurointerventional Radiology, Boston Children's Hospital  
Adriane L. Baylis\*\*, PhD; Department of Plastic Surgery, Nationwide Children's Hospital  
Francine Blei, MD; Department of Pediatrics/Hematology, Lenox Hill Hospital, Northwell Health  
Patricia E. Burrows, MD; Department of Radiology, Medical College of Wisconsin  
Sarah L. Chamlin, MD; Division of Pediatric Dermatology, The Ann and Robert H. Lurie Children's Hospital of Chicago  
Robert H. Chun, MD; Department of Otolaryngology, Medical College of Wisconsin  
Christopher P. Hess\*, MD, PhD; Departments of Radiology and Neurology, University of California, San Francisco  
Shawna Joachim\*\*; Department of Dermatology, Medical College of Wisconsin  
Katherine Johnson, DO; Department of Dermatology, Henry Ford Hospital  
Wendy Kim, DO; Division of Dermatology, Departments of Medicine and Pediatrics, Loyola University Medical Center  
Marilyn G. Liang, MD; Department of Dermatology, Harvard Medical School  
Mohit Maheshwari, MD; Department of Radiology, Medical College of Wisconsin  
Garrett N. McCoy; Department of Dermatology, Medical College of Wisconsin  
Denise W. Metry, MD; Department of Dermatology, Texas Children's Baylor  
Priya A. Monrad, MD; Department of Neurology, Medical College of Wisconsin  
Elena Pope\*, MD; Section of Paediatric Dermatology, University of Toronto and Hospital for Sick Children  
Julie Powell, MD; Department of Dermatology, University of Montreal  
Tor A. Shwayder, MD; Department of Dermatology, Henry Ford Hospital  
Dawn H. Siegel\*\*, MD; Department of Dermatology, Medical College of Wisconsin  
Megha M. Tollefson, MD; Department of Dermatology and Pediatrics, Mayo Clinic  
Sudhakar Vadivelu, DO; Department of Neurosurgery, Cincinnati Children's Hospital Medical Center  
Sean M. Lew, MD; Department of Neurosurgery, Medical College of Wisconsin  
Ilona J. Frieden\*, MD; Professor of Dermatology and Pediatrics, University of California, San Francisco  
Beth A. Drolet\*, MD; Departments of Dermatology and Pediatrics, Medical College of Wisconsin  
\* members of PHACE Syndrome Medical Advisory Board \*\* members of PHACE Syndrome Community

# Diagnosis and Care Recommendations for Children with PHACE Syndrome

- Who should be screened for PHACE syndrome
- What tests should be performed at baseline
- Review the updated diagnostic criteria
- Highlight “newer” PHACE associations
- How should children be followed once they are diagnosed with PHACE?
  - Risk Adjusted Surveillance

# Who Should Be Screened?

- Large segmental infantile hemangioma of the head
  - Face
  - Scalp
- Infants with infantile hemangioma that are:
  - Smaller or lacking typical morphology or distribution(e.g. neck, arm, chest) who have characteristic or major anomalies found in PHACE
    - Midline ventral defects
    - Coarctation of the aorta
    - Other
- Infants without skin hemangiomas with other characteristic anomalies found in PHACE
  - Major criteria
    - Cerebral vascular anomaly
    - Cardiovascular
    - Ocular anomaly
    - Midline/Ventral anomaly



# PHACE Syndrome Screening

1. Brain MRI with and without gadolinium
2. MR angiography (MRA) of the head and neck.
3. Echocardiogram
4. Ophthalmology exam

# Proposed change in Diagnostic Criteria for **Definite** PHACE Syndrome

- Hemangioma of **head including scalp** >5 cm PLUS one major criterion
  - Cerebral vascular anomaly
  - Cardiovascular
  - Ocular anomaly
  - Midline/Ventral anomaly
- **Hemangioma of the neck, upper trunk or trunk and proximal upper extremity plus two major criterion**
  - Cerebral vascular anomaly
  - Cardiovascular
  - Ocular anomaly
  - Midline/Ventral anomaly

# Proposed change in Diagnostic Criteria for Possible PHACE Syndrome

- Hemangioma of head including scalp >5 cm PLUS one minor criterion
- Hemangioma of the neck, chest or upper arm plus one major or two minor criterion
- No hemangioma but two major criterion

# Cerebrovascular Anomalies

## Risk for Stroke

- Risk Stratification
  - Low Risk
  - Intermediate Risk
  - High Risk
- Impacts surveillance recommendation
- Dr. Heyer to review in greater detail later today

# Less Frequently Recognized Associations with PHACE

- Hearing abnormalities
- Speech and Language abnormalities
- Feeding abnormalities
- Endocrine abnormalities
  - Poor growth
- Dental abnormalities
- Quality of Life/Psychosocial Impact of PHACE

## ONLINE FIRST

# Association of Hearing Loss With PHACE Syndrome

Kelly J. Duffy, PhD; Christina Runge-Samuelson, PhD; Michelle L. Bayer, BS;  
David Friedland, MD, PhD; Cecille Sulman, MD; Robert Chun, MD; Joseph E. Kerschmer, MD;  
Denise Metry, MD; Denise Adams, MD; Beth A. Drolet, MD

- Sensorineural or Conductive hearing loss
- *Hearing screening in newborn period or at time of diagnosis*
- *Repeat in “high risk” patients*
  - Intracranial hemangioma
  - Hemangiomas blocking the ear canal
  - Recurrent ear infections

Arch Dermatol. 2010;146(12):1391-1396.

# Risk of Dysphagia and Speech and Language Delay in PHACE Syndrome

Kari L. Martin, M.D.,\*,†,‡ Joan C. Arvedson, Ph.D.,‡,§ Michelle L. Bayer, M.D.,¶  
Beth A. Drolet, M.D.,‡,¶ Robert Chun, M.D.,\*\* and Dawn H. Siegel, M.D.‡,¶

*Departments of \*Dermatology and †Pediatrics, University of Missouri at Columbia, Columbia, Missouri, ‡Department of Pediatrics, Medical College of Wisconsin, Milwaukee, Wisconsin, §Department of Speech-Language Pathology and Audiology, Children's Hospital of Wisconsin, Milwaukee, Wisconsin, Departments of ¶Dermatology and \*\*Otolaryngology, Medical College of Wisconsin, Milwaukee, Wisconsin*

- At Risk Children
  - Posterior fossa brain malformations
  - Hemangiomas on the lip/ oral cavity/airway
  - Hearing loss
  - History of cardiac surgery
  - Children demonstrating feeding difficulties
- *Early referral for speech-language evaluation*
  - *At risk children no later than 24 months of age*
  - *Earlier in children who demonstrate signs or symptoms*
- *Early Intervention*

# Endocrine Abnormalities

- Thyroid abnormalities
  - Hypothyroidism
    - Abnormalities in the hypothalamus, pituitary or thyroid gland.
    - Abnormal pituitary can present as an empty sella turcica on MRI.
    - TSH and T4 might be normal in newborn period in children with PHACE who are subsequently found to have hypothyroidism
    - Neonatal screening can be diagnostic but it may miss cases with later onset
    - Repeat studies may
- Pituitary abnormalities
  - “Midline” defect
  - Poor growth/ growth hormone deficiency
  - empty or partially empty sella turcica noted on MRI but may also occur without evident brain malformations
  - Neonatal hypoglycemia can be a sign of hypopituitarism and should prompt additional endocrinologic evaluation in PHACE patient
  - Delayed pubertal onset and late-onset adrenal insufficiency
- ***Focused assessment of height, weight, and developmental milestones EXTREMELY IMPORTANT***



# Dental Anomalies

Pediatric Dermatology Vol. 31 No. 4 455–458, 2014

## Tooth Enamel Hypoplasia in PHACE Syndrome

**Yvonne E. Chiu, M.D.,\* Dawn H. Siegel, M.D.,\* Beth A. Drolet, M.D.,\* and  
Brian D. Hodgson, D.D.S.†**

*\*Departments of Dermatology and Pediatrics, Medical College of Wisconsin, Milwaukee, Wisconsin,  
†Marquette University School of Dentistry, Milwaukee, Wisconsin*

- 
- Under development of the tooth enamel
    - 28% of children
    - Leads to early dental caries
  - Associated with hemangioma within the oral cavity
  - *Intraoral hemangiomas refer to a pediatric dentist by one year of age*

# Psychosocial Impact

- Little data about PHACE specifically
  - Dr Haggstrom will discuss this in greater detail tomorrow
- Data from facial hemangiomas
  - Parental panic and disbelief
  - Stressful social interactions
    - Questions about injury, trauma, abuse
    - Public reactions leading to heightened awareness about the IH in affected families
- Larger impact on Quality of Life
  - Hemangiomas on the face
  - Larger hemangiomas
- *Primary physicians and specialists*
  - *Address parental concerns*
  - *Make appropriate referrals*

Thank You!

phace syndrome™  
community

