

Neuroimaging and Treatment of Cerebrovascular Anomalies in PHACE

Darren Orbach, MD PhD

*Neurointerventional Radiology
Boston Children's Hospital
Harvard Medical School*

PHACE

- Ageneses of arteries (absence)
- Dysplasias of arteries (abnormal size or contour)
- Stenoses of arteries (narrowing)

The Most Feared Repercussion of Cerebrovascular Anomalies:

Stroke

Stroke: Sudden onset of neurological dysfunction, due to a vascular process

Ischemic Stroke: The problem is one of blocked flow (usually due to a clot)

Hemorrhagic Stroke: The problem is one of blood bursting out of its normal vessels (arteries, veins, capillaries)

Children are not small adults!



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In adults 80% of strokes are ischemic

In children the ratio of hemorrhagic to ischemic stroke is
much closer to 50-50



Overall, Stroke in Children is Rare

WHO estimate of ischemic stroke incidence **in adults:**

~200 cases/100,000 (overall)

> 500/100,000 (\geq age 55)

Estimates of ischemic stroke incidence **in children:**

1-2/100,000

Exception: sickle cell disease patients:

~285/100,000



Pediatric Ischemic Stroke

- Neonatal / perinatal (1/4,000 live births!)
- Moyamoya
- Arterial dissection
- Cardiac abnormalities
- Clotting disorders
- Other – focal arteriopathy, sickle cell disease, vasculitis, etc.

Pediatric Hemorrhagic Stroke

- Vascular malformations (AVM, cavernous malformations)
- Arteriovenous fistulae
- Trauma
- Aneurysms

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Where does PHACE fit in?

Pediatric Hemorrhagic Stroke

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- Trauma
- Aneurysms



Examples of conditions where the underlying problem clearly impacts stroke:

- Connective tissue disorders (aneurysms, dissections)
 - Sickle cell disease, clotting disorders

The relationship in PHACE is more complex and subtle

PHACE

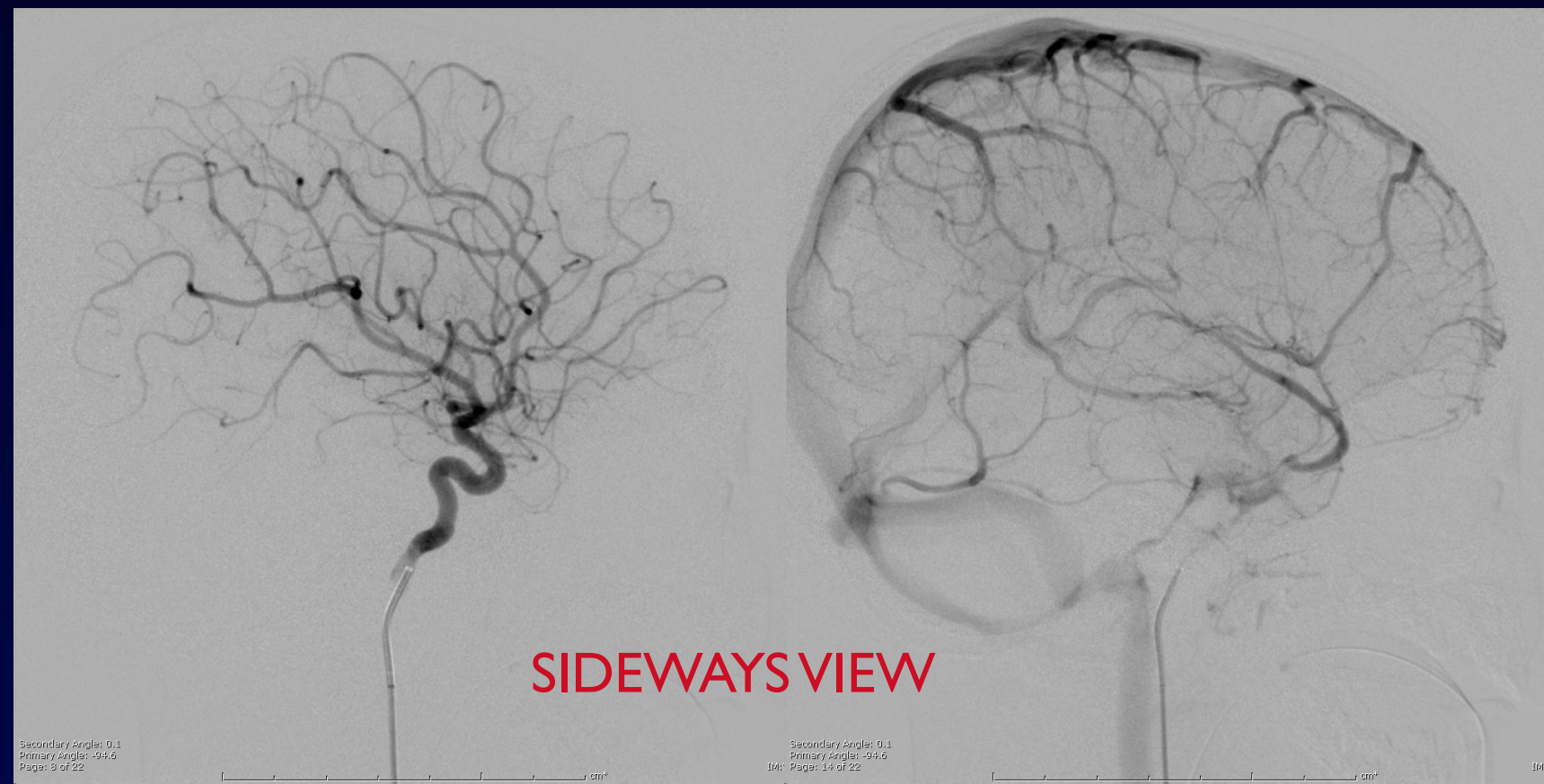
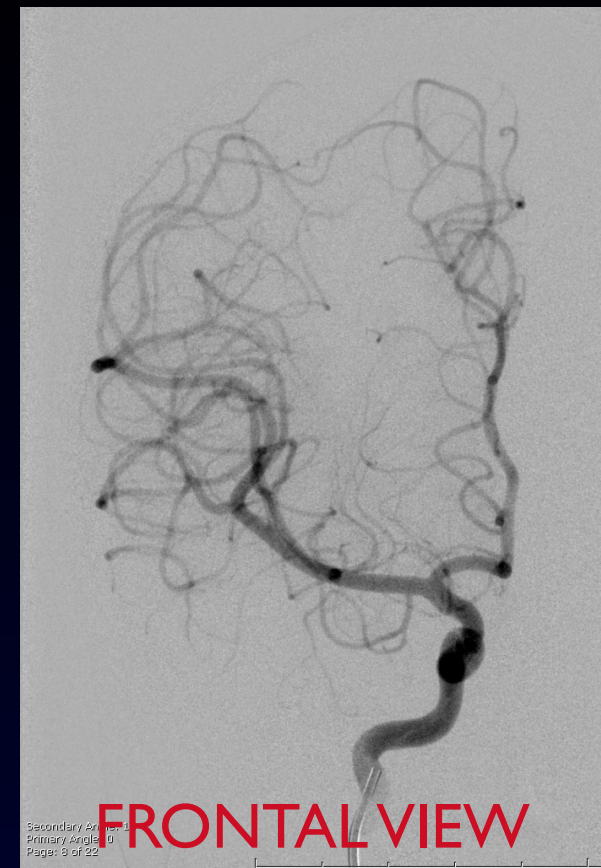
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It is still unclear whether or not there is a distinct “PHACE-related stroke,” or whether cases are due to the known categories of pediatric stroke

Overall, it seems that stroke is very rare in PHACE!

Cerebral Angiography

The mainstay of vessel neuroimaging in children is MRA!



Moyamoya

- (i) Progressive narrowing of the end of the internal carotid arteries and the first parts of the middle and anterior cerebral arteries
- (ii) Proliferation of the normally tiny deep perforating arteries



FRONTAL VIEW

SIDEWAYS VIEW

Moyamoya

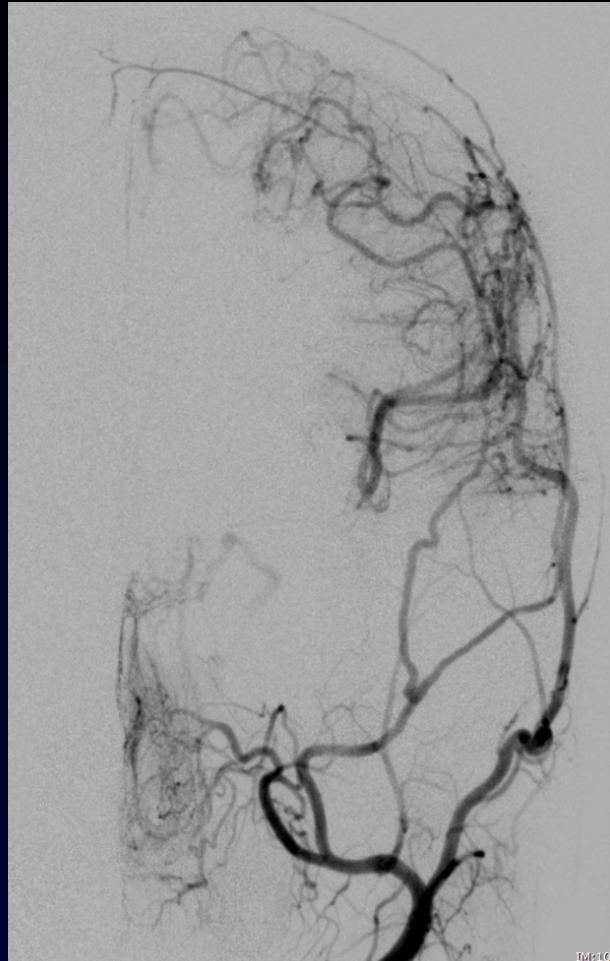
Severe and worsening headache is a common presenting symptom

Neurological symptoms follow crying, hyperventilation, coughing, straining, fever, dehydration

Stroke is multiple and recurrent, usually in the ICA territory



Moyamoya – *Pial Synangiosis*

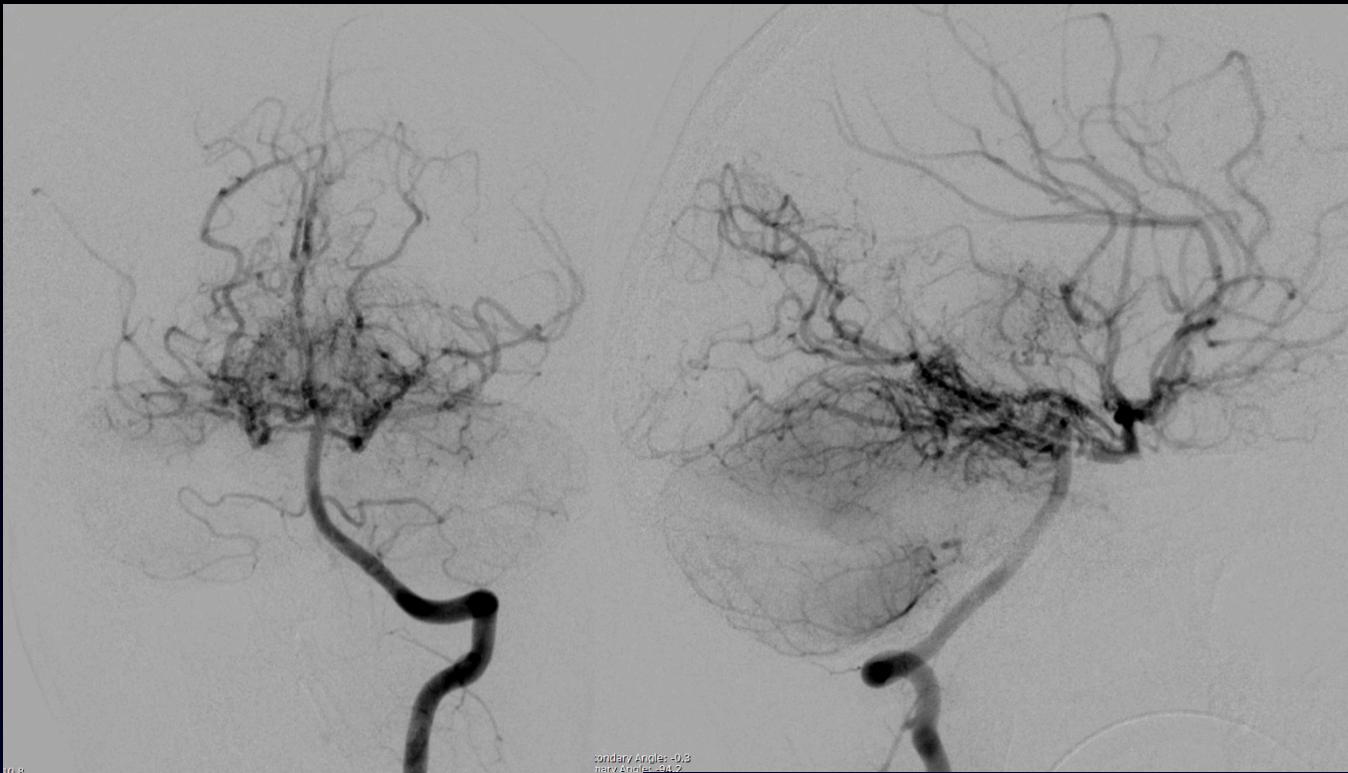


- After synangiosis, lifetime risk of stroke from moyamoya is ~4%
- Patients take daily aspirin for life

MMD in PHACE

Since 2007, out of 174 cases of treated MMD, 10 children had PHACE: **5.7%**

Note that this is different from asking what percentage of PHACE patients will develop MMD!
In every case in which the MMD affected only one side, it was on the same side as the hemangioma



Neuroimaging Recommendations

- Currently under development here at the research meeting
- Baseline MRI brain MRA brain, neck, aortic arch at age 0-1
- Follow-up imaging depends on findings: type of abnormality, location of abnormality
- Some abnormalities may not need automatic imaging follow-up in early childhood (unusual course of arteries, unusual loops or enlarged size, etc.). Later follow-up may be recommended in these cases in order to illuminate the natural history

To Summarize:

- Every child with PHACE should have a baseline vascular imaging study of the brain (MRA)
- If the baseline study is unremarkable, there is currently no reason to believe that there is increased risk of ischemic or hemorrhagic stroke. How often to continue imaging is undetermined
- The findings must be interpreted in the context of the patient:
Absence of an artery alone does not equal inadequate perfusion!
Clinical exam and other imaging signs of ischemia are crucial

To Summarize:

- If there are abnormalities that fit into broader categories of pediatric cerebrovascular disease (MMD, aneurysms), these should be followed or treated at a high-volume, specialized center, with a plan individualized for the patient
- Catheter angiography is rarely needed to diagnose vascular disease, unless there are specific questions that cannot be answered by MRA
- Our research challenge is to rigorously characterize both ischemic and hemorrhagic cerebrovascular conditions in PHACE – do they resemble the analogous conditions in children without PHACE?

Thank you



Boston
Children's
Hospital

Cerebrovascular Surgery
and Interventions Center