PHACE

- Congenital Cutaneous Hamartomas With Skeletal Muscle Differentiation Associated With LUMBAR Syndrome
- Delphi Consensus on Diagnostic Criteria for LUMBAR Syndrome
- Numerical flow experiment for assessing predictors for cerebrovascular accidents in patients with PHACES syndrome
- Non-vascular intracranial lesions in three children with PHACE association
- Pediatric Moyamoya Revascularization Perioperative Care: A Modified Delphi Study
- Scepter-Mini Balloon Assisted Coil Embolization of an Intracranial Arterial Aneurysm in a Child with PHACE Syndrome via a Persistent Trigeminal Artery
- Introduction to phacomatoses (neurocutaneous disorders) in childhood
- Fetal neuroimaging findings in PHACE syndrome: case report and review of the literature

PHACE syndrome (OMIM 606519) is a rare vascular neurocutaneous disorder, characterized by posterior fossa malformations, large cervicofacial infantile hemangiomas, arterial anomalies, coarctation of the aorta, cardiac abnormalities, and eye abnormalities.

The striking elongation and tortuosity of the distal ICA generally appeared to be a type of congenital lesion occurring early in embryogenesis as either a sporadic phenomenon or an arterial change associated with PHACE syndrome. Imaging findings revealed various mural abnormalities with a benign clinical course [1) 2)].

Diagnosis

The radiologist has an important role in the diagnosis of PHACE syndrome and in the assessment of potential complications. Investigation of infants with segmental craniofacial hemangiomas should include cranial magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) of the cerebral and cervical arteries.

Treatment

Treatment for PHACE syndrome is often tailored to the individual's specific needs and may involve a team of medical specialists, including dermatologists, cardiologists, neurologists, and ophthalmologists, among others.

The management of PHACE syndrome may include medical interventions to address specific issues, such as medication to treat hemangiomas, surgical procedures to correct vascular anomalies or heart defects, and ongoing monitoring to ensure the child's overall health and development.

Because PHACE syndrome can be complex and affect multiple organ systems, individuals with this condition typically require long-term medical care and follow-up.

A meticulous diagnostic and treatment protocol for PHACES patients with cerebrovascular anomalies within the intermediate and high-risk strata for ischemic stroke was presented by Habib et al. They also differentiate the vasculopathy associated with PHACES syndrome from moyamoya angiopathy.

Methods: Medical records and radiological imaging were reviewed. After initial magnetic resonance imaging/angiography (MRI/MRA), H215O-PET scan (baseline and Acetazolamide challenge) was performed in three patients and 6-vessel cerebral angiography was performed in two patients. Two patients with significant intracranial cerebrovascular anomalies underwent cerebral revascularization.

Results: Each patient presented with a facial hemangioma at birth and additional cerebrovascular anomalies ranging from hypoplasia to steno-occlusive changes of intracranial cerebral arteries. Additional involvement of the cardiovascular system was observed in two patients. Additional to MRI/MRA, a H215O-PET helped stratify the three patients into intermediate (n=1) and high risk groups (n=2). The high-risk group patients underwent individualized cerebral revascularization for future stroke prevention. The patient in the intermediate-risk group will be followed. Cerebrovascular angiopathy seen in all patients was typical for PHACES without moyamoya and was not progressive at follow-up.

Patients within the intermediate and high-risk strata for ischemic stroke must undergo a 6-vessel cerebral angiography and further hemodynamic evaluation to indicate the need for cerebral revascularization to prevent ischemic stroke. Non-progressive vasculopathy associated with PHACES can itself be hemodynamically relevant for neurosurgical intervention. This vasculopathy is distinct from moyamoya angiopathy, which can occur in conjunction with PHACES, resulting in concurrent progressive vasculopathy that would otherwise be absent [3)]

Outcome

Early diagnosis and comprehensive care can help improve outcomes and quality of life for those affected by PHACE syndrome. It's important for individuals with PHACE syndrome and their families to work closely with healthcare providers who are experienced in managing this rare condition.

The long-term outcome of PHACE syndrome patients is unclear; however, it seems that they are at risk for childhood stroke.

Case reports

Three pediatric patients with PHACE and non-vascular intracranial lesions [4)].

A case was referred at 24 weeks' gestation with the probable diagnosis of Dandy-Walker malformation. Prenatal sonographic examination revealed hypoplasia of the left cerebellar hemisphere, hypoplasia of the cerebellar vermis, and enlarged cisterna magna (the “tilted telephone receiver sign”). Fetal MRI at 30 weeks confirmed the findings and also revealed an ipsilateral retro cerebellar cyst communicating with the asymmetrical dilated fourth ventricle, upward displacement of the left cerebellar hemisphere, and elevation of the ipsilateral tentorium. Postnatally, a large left

facial segmental hemangioma as well as ipsilateral vascular intracranial malformations were identified, confirming the diagnosis of PHACE syndrome. A review of the literature revealed 11 reports describing 22 fetuses with prenatal imaging studies, including ours, confirming the high prevalence of specific posterior fossa abnormalities associated with PHACE syndrome.

The case ich can be identified through prenatal sonography and fetal MRI [5)]

Jagadeesan et al. report the successful treatment of a growing left posterior-communicating artery aneurysm arising from an aberrant left internal carotid artery (LICA) with balloon-assisted coiling (BAC) in a child with PHACE syndrome. They circumvented the limitations posed by the narrow caliber of the proximal LICA, by successfully navigating a coiling microcatheter from the basilar artery into the LICA via a persistent trigeminal artery. BAC was then achieved using a Scepter Mini balloon microcatheter for aneurysm neck remodeling [6)].

Brain MRI and MRA findings of a 5-year-old female patient with PHACE syndrome are presented [7)].

A 75-year-old male with PHACE anomaly, aortic anomaly, malformation of the brain, aplastic right carotid artery, and cervical vasculopathy. He presented with a transient ischemic attack with the left hemiparesis, a rare clinical presentation of the PHACE syndrome. He had an uneventful recovery and recently completed a 2-year follow-up after the superficial temporal artery to middle cerebral artery anastomosis.

PHACE syndrome should be kept in mind, even in individuals of advanced age, in the instance of a TIA, especially in situations that may involve induced hypoperfusion [8)].

Hadisurya J, Guey S, Grangeon L, Wieczorek D, Corpechot M, Schwitalla JC, Kraemer M. Moyamoya angiopathy in PHACE syndrome not associated with RNF213 variants. Childs Nerv Syst. 2019 Apr 29. doi: 10.1007/s00381-019-04145-9. [Epub ahead of print] PubMed PMID: 31037424.

A child with a throat hemangioma, vascular malformations, cognitive delay, and other anomalies illustrate the neuroimaging found in this syndrome [9)].

Test

Here's a multiple-choice test about PHACE syndrome based on the provided information:

What does PHACE syndrome stand for? a) Pediatric Hemangioma and Cardiac Abnormalities Syndrome b) Posterior Fossa Brain Malformation and Arterial Anomalies Syndrome c) PHACE syndrome is not an acronym d) Posterior Fossa, Hemangiomas, Arterial Anomalies, Cardiac

Abnormalities, Eye Abnormalities Syndrome

Which of the following is a characteristic feature of PHACE syndrome? a) Kidney abnormalities b) Respiratory issues c) Large cervicofacial infantile hemangiomas d) Bone fractures

What role do radiologists play in the diagnosis of PHACE syndrome? a) Radiologists have no role in the diagnosis of PHACE syndrome b) Radiologists conduct genetic testing c) Radiologists perform cranial magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) d) Radiologists administer medications for treatment

How is the treatment for PHACE syndrome typically managed? a) Exclusively by dermatologists b) With surgical procedures for heart defects only c) Tailored to the individual's specific needs and involves a team of medical specialists d) Treated with over-the-counter medications

What is one potential long-term complication associated with PHACE syndrome? a) Allergic reactions to medications b) Childhood stroke c) Respiratory infections d) Hearing loss

How can the vasculopathy associated with PHACE syndrome be distinguished from moyamoya angiopathy? a) By performing an electrocardiogram (ECG) b) By evaluating skin abnormalities c) Through medical history alone d) By performing a 6-vessel cerebral angiography

What imaging technique is recommended for investigating infants with segmental craniofacial hemangiomas suspected of having PHACE syndrome? a) X-rays b) CT scans c) Cranial magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) d) Ultrasound

What is one finding that can be observed in patients with PHACE syndrome based on brain MRI and MRA? a) Hypertension b) Liver abnormalities c) Hypoplasia of cerebellar vermis d) Skin rashes

What is the primary purpose of cerebral revascularization in patients with PHACE syndrome? a) To treat infantile hemangiomas b) To prevent ischemic stroke c) To correct eye abnormalities d) To improve cardiac function

In which case should PHACE syndrome be considered, even in individuals of advanced age? a) When there is a history of diabetes b) When experiencing joint pain c) In the instance of a transient ischemic attack (TIA) d) When there is a history of respiratory infections

Answers

d) Posterior Fossa, Hemangiomas, Arterial Anomalies, Cardiac Abnormalities, Eye Abnormalities Syndrome c) Large cervicofacial infantile hemangiomas c) Radiologists perform cranial magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) c) Tailored to the individual's specific needs and involves a team of medical specialists b) Childhood stroke d) By performing a 6-vessel cerebral angiography c) Cranial magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) c) Hypoplasia of cerebellar vermis b) To prevent ischemic stroke c) In the instance of a transient ischemic attack (TIA)

1)

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