

New PHACE Syndrome Study: Longitudinal Study of Neurologic, Cognitive and Radiologic Outcomes in PHACE Syndrome.

What is PHACE syndrome?

A small group of patients with skin hemangiomas (non-cancerous growth of blood vessels appearing as a type of birthmark) on the head and neck may also have additional associated disorders in what is called PHACE Syndrome. **PHACE** refers to **P**osterior fossa anomalies (a structural brain abnormality); **H**emangioma (growth of blood vessels appearing on the skin); **A**rterial abnormalities (arteries are blood vessels in your body that carry blood away from your heart); **C**ardiac (heart) abnormalities; and abnormalities of the **E**ye. Your child may have one or several of the abnormalities listed above as part of the PHACE syndrome.

Why is this study important?

While it is known that children with PHACE syndrome may have abnormalities of the blood vessels in the brain or structural brain abnormalities, it is not understood what the significance of these abnormalities is or how this affects children as they get older. As a result, it is difficult for physicians to counsel parents of infants with PHACE syndrome regarding future expectations or problems that may be encountered in regards to development. This project is the first study to look at specific areas of development in children with PHACE syndrome through neurologic, psychological and cognitive evaluations. The data used from this study will be used in the development of standardized testing to establish clinical guidelines for the management of children with PHACE syndrome. In addition, we believe that the information gained in this study will better characterize PHACE syndrome, and establish guidelines for diagnostic neuroimaging of at-risk infants.

To participate in this trial:

- Your child must be diagnosed with definite or probable PHACE syndrome (according to the 2009 diagnostic criteria)
- Your child must be 4, 5 or 6 years of age.
- You and your child must be available to travel to Children's Hospital of Wisconsin (CHW) for non-invasive evaluations with Beth Drolet, MD and her research team over a one to two day period.

How many patients will be enrolled in the study?

About 30 children will be enrolled in this study.

What will happen at the study visit?

- This study will look at specific areas of your child's development.
- Your child will have a standard neurological exam with a pediatric neurologist that includes:
 1. **Cognitive exam:** your child may be asked to do simple tasks, such as count from 1 to 5.
 2. **Eye exam:** your child may be asked to identify shapes on a chart such as a dog, heart, or a ball.
 3. **Motor exam:** your child's muscle strength will be tested, for example they may be asked to pick up a toy and hand it to you or shake your hand.
 4. **Sensory exam:** the examiner may touch your child's arm or foot and see if they can feel the touch.
 5. **Cerebellar exam:** your child may be asked to do a task like touch their nose.
- Your child will have a neurocognitive evaluation with a pediatric psychologist. This will include testing of your child's memory and recognition using puzzles and memory type games, for example. This will take 2 hours.
- You will be asked questions about pregnancy, family history and your child's medical history. This will take 60 minutes. These questions are used to determine the possible genetic links to PHACE syndrome.
- You and your child should expect to spend 4 to 5 hours at CHW.
- You will not be charged for the evaluations.
- You will be reimbursed for travel expenses up to \$500.
- No invasive testing will be done.
- No drugs will be given in this study.
- No experimental techniques will be part of this study.

What happens after the study?

You will receive a written report on all the testing that was performed on the day of your study visit. The data will be collected and compared to other features of PHACE syndrome such as sex, presence of heart or aortic arch disease, and cerebrovascular disease.

For more information about the study, please contact the research coordinator:

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For more information on PHACE syndrome or hemangiomas go to www.chw.org