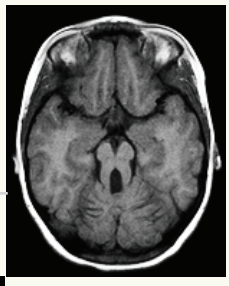
UNIVERSITY of WASHINGTON

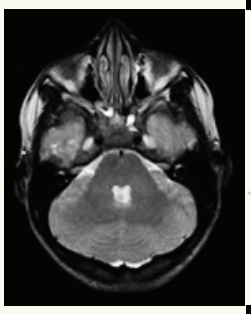
Hindbrain Malformation Research Program

Our Research

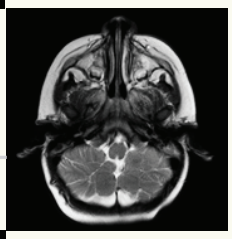
We study the biology of hindbrain malformations and neurodevelopmental disorders such as Joubert, Poretti-Boltshauser, and Chudley-McCullough syndromes, rhombencephalosynapsis, pontine tegmental cap dysplasia, and tubulinopathies to improve the lives of families affected by these conditions and advance the understanding of brain development. Identifying the causes of these disorders may provide better diagnostic and prognostic information to families and lead to better treat-ments in the future.



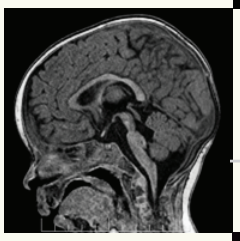
Joubert syndrome



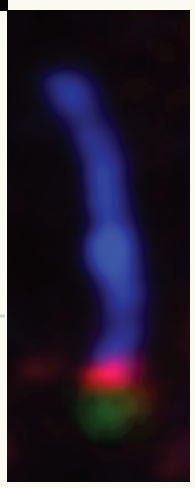
Rhombencephalosynapsis



Chudley-McCullough syndrome



Pontine tegmental cap dysplasia



Primary cilium in a neural progenitor cell labeled with ARL13B (blue), CC2D2A (red) and gamma tubulin (green).

Get Involved!

Participation includes four parts:

MRI review. To determine eligibility, we analyze brain imaging. We may also review prenatal images, including ultrasounds and fetal MRIs, as well as X-rays and CT scans.

Consent process. We will explain the research study and answer your questions about our consent forms before you agree to participate.

DNA collection. Different types of samples provide us with different information. We primarily ask for blood or saliva, but we will sometimes ask permission for a skin sample.

Medical information collection. We will conduct a brief interview and request medical records.

Please contact us for more information or to participate in our research.

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