



# Midbrain/Hindbrain Malformations and Hydrocephalus: Understanding the Causes, Consequences and Gaps in Understanding

Midbrain/Hindbrain Malformations and Hydrocephalus Workshop 2014

Neuroscience Center  
6001 Executive Blvd.  
Room C/D  
Bethesda, MD

## AGENDA

### MONDAY, June 23<sup>rd</sup>

- 7:30–8:00 am**      **Registration**
- 8:00 am**            **Opening and Welcome:**  
8:00–8:10 am  
                         Story Landis, Director, NINDS
- 8:10–8:20 am  
                         Jill Morris, Program Director, NINDS, *Overview of NIH efforts*
- 8:20 am**            **Session 1: Classifications: Purpose and Problems (Chair: Jim Barkovich)**
- 8:20–8:50 am  
                         Dan Doherty, University of Washington: *Advances in midbrain-hindbrain malformation diagnosis*
- 8:50–9:20 am  
                         Dave Limbrick, Washington University School of Medicine: *Hydrocephalus: A Clinician's Perspective*
- 9:20–9:50 am  
                         Jim Barkovich, University of California, San Francisco: *MBHB Malformations: a classification*
- 9:50–10:05 am**      **BREAK**
- 10:05–10:35 am  
                         Francis Brunelle and Nathalie Boddaert, Necker Hospital: *Midbrain-hindbrain malformation. A practical genetic imaging algorithm: from MRI to genes*

10:35–11:05 am

Charles Raybaud, The Hospital for Sick Children: *Fetal hydrocephalus with mid/hindbrain malformation: what do we understand?*

11:05–11:35 am

Hannah Tully, Seattle Children's Research Institute: *A new approach to the classification of developmental hydrocephalus*

**11:35 – 1:15 pm**

**WORKING LUNCH: PANEL DISCUSSION**

**Panel Discussion of Objective 1:**

- Classifications and Diagnosis: Purpose and Problems
  - a. Pre-natal malformation detection: Is it predictive?
  - b. Structural features: Correlation with natural history, outcomes

**1:15 pm**

**Session 2: Genetics of Midbrain/Hindbrain Malformations and Congenital Hydrocephalus: common biology? (Chair: Bill Dobyns)**

1:15-1:45 pm

Bill Dobyns, Seattle Children's Research Institute: *A new approach to the genetics of developmental hydrocephalus*

1:45–2:15 pm

Naiara Akizu, University of California, San Diego: *Unveiling mechanism and treatment of cerebellar disorders from disease gene discovery*

2:15–2:45 pm

Colin Johnson, University of Leeds: *Insights into the complex disease mechanisms of severe ciliopathies: from genes to mouse models*

**2:45 –3:00 pm**

**BREAK**

3:00–3:30 pm

Viktor Chizhikov, The University of Tennessee Health Science Center: *Misspecification of cerebellar neurons into brain stem fates leads to cerebellar agenesis in the absence of Ptf1a function*

3:30–4:00 pm

Tamara Caspary, Emory University: *Interpreting signaling through the primary cilium*

4:00–4:30 pm

Allison Ashley-Koch, Duke University School of Medicine: *Genetics of Chiari Malformations: Current findings*

4:30–5:00 pm

Russell J. Ferland, Albany Medical College: *Mutations in CSPP1 cause primary cilia defects and Joubert syndrome*

5:00 – 6:00 pm

**PANEL DISCUSSION**

**Discussion of Objective 2:**

- Genetics of Midbrain/Hindbrain Malformations and Hydrocephalus: Common biology?

6:30 pm

**Dinner and Presentations by the Sponsors**

Dinner is sponsored by the Hydrocephalus Association, Chiari & Syringomyelia Foundation and the Dandy Walker Alliance

**TUESDAY, June 24th:**

8:00 am

**Session 3: Animal Models of Midbrain/Hindbrain Malformations and Hydrocephalus: What can we learn? (Chair: Kathleen J. Millen)**

8:00–8:30 am

Roy Sillitoe, Baylor College of Medicine: *Mapping cerebellar circuits: development and dysfunction*

8:30–9:00 am

Alexandra Joyner, Sloan-Kettering Institute: *Regulating the production of input neurons and target fields to form normal cerebellar circuits*

9:00–9:30 am

Eva Anton, University of North Carolina at Chapel Hill School of Medicine: *Arl13b, Primary Cilia, and the Formation of Cerebral Cortex*

9:30–9:45 am

**BREAK**

9:45–10:15 am

Kathleen Millen, U of Washington: *Head mesenchyme influences very early choroid plexus and cerebellar development*

10:15 – 10:45 am

Tim Vogel, Cincinnati Children's Hospital: *Ciliopathy mouse models to study the genesis of hydrocephalus*

10:45 – 11:15 am

Jennifer Schmidt, University of Illinois at Chicago: *Abnormal ciliary microtubule organization and juvenile hydrocephalus in *Jhy*<sup>lacZ</sup> mice*

11:15 – 11:45 am

James P. (Pat) McAllister, University of Utah: *Critical comparisons between genetic and acquired models of hydrocephalus*

**11:45 – 1:00 pm**

**WORKING LUNCH: PANEL DISCUSSION**

**Panel Discussion of Objective 3:**

- Animal models of Midbrain/Hindbrain Malformations and Hydrocephalus: What can we learn?

**1:00 pm**

**Session 4: New Models for Thinking about Midbrain/Hindbrain Malformations and Hydrocephalus and Potential Treatments (Chair: Dave Limbrick)**

1:00–1:30 pm

Bonnie Blazer-Yost, Indiana University-Purdue University: *TRP channels in hydrocephalus*

1:30–2:00 pm

Jerold Chun, Scripps Research Institute: *Lysophospholipid signaling in post-hemorrhagic hydrocephalus*

2:00-2:30 pm

Dave Limbrick, Washington University School of Medicine: *CSF Markers of Hydrocephalus: Progress Towards Improving Clinical Management and Outcomes*

**2:30-2:45 pm**

**BREAK**

2:45-3:15 pm

Anthony Wynshaw-Boris, Case Western University School of Medicine: *The Wnt/PCP pathway, ependymal cilia polarity and hydrocephalus*

3:15-3:45 pm

Gregory Heuer, The Children's Hospital of Philadelphia: *Impact of Fetal Myelomeningocele Closure on Chiari Malformation and Hydrocephalus*

**3:45-4:45 pm**

**FINAL PANEL DISCUSSION**

**Discussion of Mission Aim 4:**

- New Models for Thinking about Midbrain/Hindbrain Malformations and Hydrocephalus and Potential Treatments

**4:45-5:30 pm**

**SUMMARY AND MOVING FORWARD**