



JOHNS HOPKINS  
MEDICINE  
CONTINUING MEDICAL EDUCATION

*The National Institute of Neurological  
Disease and Stroke,  
The Office of Rare Diseases,\*  
The Johns Hopkins University School  
of Medicine  
and The PND Association  
Present*

**Medical Management of Pediatric  
Neurotransmitter Disorders:  
A Multidisciplinary Approach**

**Friday, July 18 to  
Saturday, July 19, 2008**

**Hyatt Dulles at Washington  
Dulles International Airport  
Herndon, VA**

## Program Description and Objectives

Although much progress has been made in the field of Pediatric Neurotransmitter Diseases (PND's) much remains to be achieved in terms of diagnosis, understanding and treatment of these disorders. The conference will bring together scientists and clinicians to discuss current and potential treatments for PND's and to arrive at practical guidelines for multidisciplinary treatment of the various PND's.

The conference format features formal presentations by outstanding clinicians and researchers in the field, question and answer sessions, panel discussions and open dialogue. The conference format is designed to encourage interaction and collaboration between speakers and attendees concerning the best directions for clinical practice and research of PND's. Families will have an opportunity to meet "face to face" with others dealing with the same disorders and the associated challenges.

After attending this conference participants will be able to:

- Apply the latest scientific information toward the diagnosis and clinical management of PND's as measured by earlier diagnosis, increased incidence and improved patient outcomes.
- Recognize the rationale to consider and adopt a multidisciplinary treatment approach to PND's as measured by enhanced patient outcomes after diagnosis.
- Relate information gained to generate new research, new approaches and/or new collaborations on behalf of PND's, as measured by increased research, new treatments and increased awareness.

## Who Should Attend

This activity is intended for :

- Child Neurologists
- Metabolic Specialists
- Geneticists
- Health Care Professionals involved with neurological diseases
- Scientists
- Families and Caregivers

## Accreditation Statement

This activity has been planned and implemented in accordance with the Essential Areas and policies of the Accreditation Council for Continuing Medical Education through the joint sponsorship of The Johns Hopkins University School of Medicine and The National Institutes of Health. The Johns Hopkins University School of Medicine is accredited by the ACCME to provide continuing medical education for physicians.

## Credit Designation Statement

The Johns Hopkins University School of Medicine designates this educational activity for a maximum of 10.75 *AMA PRA Category 1 Credit(s)*<sup>™</sup>. Physicians should only claim credit commensurate with the extent of their participation in the activity.

## Policy on Speaker and Provider Disclosure

It is the policy of The Johns Hopkins University School of Medicine that the speaker and provider disclose real or apparent conflicts of interest relating to the topics of this educational activity, and also disclose discussions of unlabeled/unapproved uses of drugs or devices during their presentation (s). The Johns Hopkins University School of Medicine OCME has established policies in place that will identify and resolve all conflicts of interest prior to this educational activity. Detailed disclosure will be made in the activity handout materials.

## Session Chairs

**Darryl C. De Vivo MD**, Sidney Carter Professor of Neurology, Professor of Pediatrics, Director Pediatric Neurology Emeritus Columbia University Medical Center, New York NY

**K. Michael Gibson PhD, FACMG**, Director Biochemical Genetics, Childrens Hospital Pittsburgh, Pittsburgh PA

**Katherine Swoboda MD, FACMG**, Associate Professor Neurology, University of Utah School of Medicine, Salt Lake City UT

**Dan Tagle PhD**, Program Director NIH/Neuroscience Center, Bethesda MD

## Presenters

**Sally Evans MD**, Division Chief Physical Medicine and Rehabilitation, Medical Director Rehabilitation, Children's National Medical Center, Washington DC

**K. Michael Gibson PhD, FACMG**, Director Biochemical Genetics Laboratory, Childrens Hospital of Pittsburgh, Pittsburgh PA

**Ellen J. Hess PhD**, Associate Professor Neurology and Neuroscience, The John Hopkins University School of Medicine, Baltimore MD

**Friederike Hoerster MD**, Department Child & Youth Medicine, University of Heidelberg, Germany

**Keith Hyland PhD**, Director Department of Neurochemistry, Medical Neurogenetics LLC, Atlanta GA

**Un Jung Kang MD**, Associate Professor of Neurology and Pharmacological and Physiological Sciences, Co Director Parkinson's Disease and Movement Disorders Center University of Chicago, Chicago IL

**Sue Kratz, OTR CST**, Director Special Therapies, Waukesha WI

**Joanne Kurtzburg, MD**, Chief Department Pediatrics Division of Blood and Marrow Transplantation, Duke University, Durham NC

**Nicola Longo, MD PhD**, Medical Genetics/Pediatrics, Primary Childrens Medical Center, Salt Lake City UT

**Phillip Pearl MD**, Associate Professor of Pediatrics and Neurology, Children's National Medical Center, Washington DC

**Roser Pons MD** Pediatric Neurologist Movement Disorders, Mitera Hospital Athens, Greece

**Michael Rotstein MD**, Fellow Pediatric Movement Disorders, Columbia University Medical Center, New York NY

**Jerold Vitek MD PhD**, Director Neuromodulation Research Center, Cleveland Clinic, Cleveland OH

# Program Agenda

## Thursday July 17<sup>th</sup>, 2008

- 7:00 - 8:30 PM Welcome Reception  
8:30 - 10:00 PM PND Board of Directors and Medical & Scientific Advisory Board Meeting

## Friday July 18<sup>th</sup>, 2008: Day One

- 7:00 - 8:00 AM Continental Breakfast/Registration

### Morning Theme: Phenotypes and Therapy, Chair: Darryl De Vivo MD

- 8:00 - 8:15 AM Welcome and Opening Remarks, Darryl De Vivo MD, Dan Tagle PhD  
8:15 - 9:00 AM The Phenotypic Spectrum of Pediatric Neurotransmitter Diseases and Infantile Parkinsonism, Roser Pons MD  
9:00 - 9:45 AM Tetrahydrobiopterin Deficiency in Pediatric Neurotransmitter Disorders, Sepiapterin Reductase Deficiency, Nicola Longo MD  
9:45 - 10:00 AM Questions and Discussion  
10:00 - 10:30 AM Coffee/Break  
10:30 - 11:15 AM Treatment Approaches in Gamma-Hydroxybutyric Aciduria: Lessons From Mice and Men and the NIH Protocols, Phillip Pearl MD  
11:15 - 12:00 PM Treatment Modalities for L-Aromatic Amino Acid Decarboxylase Deficiency-An International Study Group, Friederike Hoerster MD  
12:00 - 12:15 PM Questions and Discussion  
12:15 - 1:00 PM Box Lunch

### Afternoon Theme: Non-Pharmacological Intervention and Alternatives Chair: K. Michael Gibson PhD,FACMG

- 1:00 - 1:45 PM Novel Treatment Approaches for Disorders of Neurotransmission-Umbilical Stem Cells, and Gene Therapy Update, Joanne Kurtzberg MD and Un Jung Kang MD  
1:45 - 2:30 PM Sensory Integration Intervention in PNDs, Sue Kratz OTR CST  
2:30 - 3:00 PM Coffee/Break  
3:00 - 3:45 PM Physical and Occupational Intervention for Pediatric Neurotransmitter Disorders-Goals and Objectives, Sally Evans MD  
3:45 - 4:30 PM Deep Brain Stimulation as Adjuvant Therapy in Pediatric Neurotransmitter Disorders, Jerrold Vitek MD, PhD  
4:30 - 5:00 PM Roundtable Discussion-Summary of Day, Darryl DeVivo MD  
5:00 PM Adjourn  
6:30 PM Dinner

## Saturday July 19<sup>th</sup>, 2008: Day Two

- 7:00 - 8:00 AM Continental Breakfast

### Morning Theme: Research Updates and Future Directions Chair: Kathryn Swoboda MD

- 8:00 - 10:10 AM Short presentations (20 min) Topic: Updates on Disorders of Neurotransmission  
8:00 - 8:20 AM Sepiapterin Reductase Deficiency and the Pediatric Neurotransmitter Disease Database, Nicola Longo MD  
8:20 - 8:40 AM Mouse Model of Tyrosine Hydroxylase:Clinical Variability in TH Deficiency as a Direct Consequence of the Influence of the Genetic Mutation on the Functions of the TH Enzyme, Ellen J. Hess PhD  
8:40 - 9:00 AM Questions and Discussion  
9:00 - 9:30 AM Coffee/Break  
9:30 - 9:50 AM Comprehensive Metabolite, Enzyme and Molecular Diagnostics for PND's, Keith Hyland PhD  
9:50 - 10:10AM Hepatocyte Repopulation in Gamma-Hydroxybutyric Aciduria, K. Michael Gibson PhD, FACMG  
10:10 - 10:30AM Combined Research/Clinical Fellowship for PND's, Michael Rotstein MD  
10:30 - 10:45 AM Break  
10:45 - 11:15 AM The Five Year Roadmap for the PND Association: PND Board of Directors  
11:15 - 12:15 PM Roundtable, Questions and Answers with All Speakers from All Days.  
12:15 - 12:30 PM Closing Remarks, Darryl De Vivo MD, Dan Tagle PhD

### Family Program Schedule

- 12:30 - 1:15 PM Bag Lunch for families sponsored by PND Association  
1:15 - 1:30 PM Welcome/ Board Introductions, Nancy Speller President & Brad Hoffman Vice President/Treasurer  
1:30 - 1:45 PM Hope for Tomorrow Begins Today, PND Fundraising Committee  
1:45 - 3:00 PM Round Table Discussions Dopamine Related Diseases/GABA Related Diseases ALL  
3:00 - 3:15 PM Break  
3:15 - 4:30 PM Continued Round Table Discussions Dopamine Related Diseases /GABA Related Diseases ALL  
4:30-4:45 PM Closing Remarks/Adjourn

**\* Funding for this conference was made possible (in part) by NS60363 from National Institute of Neurological Disorders and Stroke (NINDS) and the Office of Rare Disorders (ORD). The views expressed in written conference materials or publications and by the speakers and moderators do not necessarily reflect the official policies of the Department of Health and Human Services; nor does mention of trade names, commercial practices, or organizations imply endorsement by the U.S. Government.**

**The Johns Hopkins University School of Medicine takes responsibility for the content, quality and scientific integrity of the CME activity.**

This agenda was prepared March, 2008 and is subject to change by conference time. For program updates, please visit [www.pndassoc.org](http://www.pndassoc.org).

*The Pediatric Neurotransmitter Disease Association is a non-profit organization dedicated to helping children and families affected by Pediatric Neurotransmitter Diseases.*



Pediatric Neurotransmitter  
Disease Association

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## Registration Information

*Registration is limited so register today*

Complete the form enclosed in this packet. Fax or mail the completed form with check or money order made payable to the PND Association in US Currency to:

The PND Association  
28 Prescott Place  
Old Bethpage NY 11804  
Phone/Fax: 516 249-4948  
Email: PND@PNDAssoc.org  
ATTN: Symposium

***Registration can also be completed online (charge cards accepted) at [www.pndassoc.org](http://www.pndassoc.org)***

### **Registration Fee**

Registration fees include symposium, continental breakfast, coffee breaks, lunch and Friday dinner. Payable to PND Association in US Currency.

- \$125.00 Professionals
- \$75 per Family
- No Fee - If you are a NIH employee or affiliate there is no fee. Please provide NIH contact information on registration form.

### **Hotel Information; RESERVE TODAY**

Rooms are limited. Please call the Dulles Hyatt at Washington Dulles Airport Herndon, Virginia 703 713-1234. Mention the PND Association. Reduced rates are available.

### **Special Accommodations**

If you require special accommodations to participate in this activity please call the hotel at 703 713-1234 during the business hours of 9:00am–5:00pm at least 14 days prior to the event. If you require special accommodations to participate in this activity, please list your requests on the registration form.

### **Child Care**

Although formal childcare will not be offered at the conference, a childcare room will be adjacent to meeting rooms for easy access. One parent/caregiver must accompany the child. Contact Carolyn Hoffman at 414-646-5133 or [choffman@spsinet.com](mailto:choffman@spsinet.com) with questions regarding childcare.

### **Additional Information:**

All registrations by mail will be confirmed by email. Conference material will be distributed on site the day the conference begins. For up to date information visit the PND website at [www.pndassoc.org](http://www.pndassoc.org) . For all other inquiries contact: Stephanie Martin, PND Operations Assistant at [smarti93@optonline.net](mailto:smarti93@optonline.net) or call 516 249-4948.