42nd Annual Meeting of the Child Neurology Society

Austin, Texas

October 30 - November 2, 2013

E. Steve Roach, MD; President, CNS
Mustafa Sahin, MD, PhD; Chair, CNS Scientific Selection and Program Planning Committee

This activity has been planned and implemented in accordance with the Essential Areas and Policies of the Accreditation Council for Continuing Medical Education (ACCME) through the joint sponsorship of Nationwide Children's Hospital and the Child Neurology Society. Nationwide Children's Hospital is accredited by the ACCME to provide continuing medical education for physicians.

NCH designates this educational activity for a maximum of 26.0 AMA PRA Category 1 credits™. Physicians should only claim credit commensurate with the extent of their participation in the activity.

**PROGRAM**

**Wednesday, October 30**

**7:30 AM – 5:00 PM**  
Organizer: Bernard L. Maria, MD, MBA; Georgia Regents University, Augusta, GA

**Symposium I: Neurobiology of Disease in Children: Mitochondrial Disease**

7:30 AM – 7:40 AM  
Opening Comments  
Bernard L. Maria, MD, MBA, Principal Investigator

7:40 AM – 9:40 AM  
SESSION I: CLINICAL ASPECTS  
Co-Director and Moderator: Bruce H. Cohen, MD; Akron Children's Hospital, Akron, OH

7:40 AM – 8:05 AM  
Natural Histories and Classification of Mitochondrial Disease  
Eric Schon, PhD; Columbia University, New York, NY

8:05 AM – 8:30 AM  
Diagnosis and Treatment of Mitochondrial Diseases  
Bruce H. Cohen, MD

8:30 AM – 8:55 AM  
Mitochondrial DNA Disorders  
Patrick Chinnery, PhD/MRCPath/MRCP; New Castle upon Tyne, UK

8:55 AM – 9:20 AM  
POLG: Primary and Secondary Disorders  
Robert K. Naviaux, MD, PhD; University of California, San Diego, San Diego, CA

9:20 AM – 9:40 AM  
Question and Answer Session

9:40 AM – 9:55 AM  
Coffee Break

9:55 AM – 11:30 AM  
SESSION II: Molecular Mechanisms  
Co-Director and Moderator: William Copeland, PhD; NIEHS, Research Triangle Park, NC

9:55 AM – 10:20 AM  
Defects of Mitochondrial DNA Replication  
William Copeland, PhD

10:20 AM – 10:45 AM  
Mitochondrial Dynamics and Parkinsons  
Richard Youle, PhD; NINDS, Bethesda, MD

10:45 AM – 11:10 AM  
Systems Biology Approach to Mitochondrial Disease  
Vamsi Mootha, MD; Harvard Medical School, Boston, MA

11:10 AM – 11:30 AM  
Question and Answer Session

11:30 AM – 1:00 PM  
Lunch and Presentation by the United Mitochondrial Disease Foundation

1:00 PM – 2:40 PM  
SESSION III: TRANSLATIONAL SCIENCE AND CLINICAL FRONTIERS  
Co-Director and Moderator: Greg Enns, PhD; Stanford University School of Medicine, Stanford, CA

1:00 PM – 1:25 PM  
Gene Therapy and Enzyme Replacement  
Michio Hirano, MD; Columbia University Medical Center, New York, NY
1:25 PM – 1:50 PM  
Exercise Therapies in Attenuating Mitochondrial Disease  
Mark Tarnopolsky, MD, PhD, FRCP; McMaster University, Hamilton, ON

1:50 PM – 2:15 PM  
Clinical Trials: Edison Pharmaceuticals, EPI-743  
Greg Enns, PhD

2:15 PM – 2:40 PM  
Occupational, Environmental and Therapeutically Acquired Mitochondrial Disorders  
Kendall Wallace, PhD; University of Minnesota-Duluth, Duluth, MN

2:40 PM – 3:00 PM  
Question and Answer Session

3:00 PM – 3:15 PM  
In-Room Coffee Break

3:15 PM – 3:45 PM  
Executive Summary of the Day  
Bruce H. Cohen, MD and William Copeland, PhD

3:45 PM – 4:45 PM  
SESSION IV: FUTURE DIRECTIONS  
PANEL DISCUSSION  
Moderator: Doug Wallace, PhD; Children's Hospital of Philadelphia, Philadelphia, PA  
PANELISTS:  
Amy Goldstein, MD; Children's Hospital of Pittsburgh, Pittsburgh, PA  
Andrea Gropman, MD; Children's National Medical Center, Washington, DC  
Richard Haas, MD; University of California, San Diego, San Diego, CA  
Sumit Parikh, MD; Cleveland Clinic, Cleveland, OH  
Russ Saneto, DO, PhD; Seattle Children's Hospital, Seattle, WA

4:45 PM – 5:00 PM  
Closing Comments and Thanks  
Bernard L. Maria, MD, MBA

Thursday, October 31

7:00 – 8:15 AM  
CONTINENTAL BREAKFAST AND SEMINARS

Breakfast Seminar 1: Shifting Models of Health Care Delivery and the Child Neurologist: The Sky is Falling  
Organizer: Mark Mintz, MD; The Center for Neurological and Neurodevelopmental Health (CNNH) and the Clinical Research Center of New Jersey (CRCNJ), Gibbsboro, NJ  
Mark Mintz, MD  
Shifts in Reimbursements: Global Payments, Accountable Care Organizations and Medical Homes  
Bruce Cohen, MD; Akron Children's Hospital, Akron, OH  
Hospital-Employed Physicians: Impact of Diminishing Hospital Admissions  
Ram Kairam, MD; Columbia University Medical Center, New York, NY  
Innovative Models of Specialty Care Delivery  
Mark Mintz, MD

Breakfast Seminar 2: Tourette Syndrome - Novel Treatments at the Ends of the Spectrum  
Organizer: Leon Dure, MD; University of Alabama at Birmingham, Birmingham, AL  
Leon Dure, MD  
Medical and Surgical Approaches to TS  
Leon Dure, MD  
Examining the Outcomes Associated with Behavior Therapy for TS  
Douglas Woods, PhD; Texas A&M University  
Practical Application of Cognitive/Behavioral Therapy for TS: an OT Perspective  
Jan Rowe, DrOT; Children's Hospital of Alabama, Birmingham, AL

Breakfast Seminar 3: Refractory Status Epilepticus: An Update  
Organizer: James J. Riviello, Jr., MD; Columbia University Medical Center/Children's Hospital of New York, New York, NY  
James J. Riviello, Jr., MD  
Refractory Status Epilepticus: Definition, stages, etiologies, and therapeutic implications

8:00 AM – 4:30 PM  
Association of Child Neurology Nurses

2:00 PM – 5:00 PM  
Professors of Child Neurology

6:00 PM – 8:00 PM  
OPENING RECEPTION  
Austin Hilton Hotel, 6th Floor  
Supported by Texas Children's Hospital, Houston, TX

8:00 – 10:00 PM  
SIG Meetings (including Movement Disorders)
Standard and alternative treatments for refractory status epilepticus and the importance of a specific protocol.
Nicolas S. Abend, MD; University of Pennsylvania, Philadelphia, PA

8:45 AM – 8:50 AM
Association of Child Neurology Nurses Claire Chee Award for Excellence

- Cheryl Fischer, PNP; Memorial Sloan Kettering Cancer Center, New York, NY

8:50 AM – 9:00 AM
CNS Lifetime Achievement Awards:
- Arthur Rose, MD; SUNY Downstate, Brooklyn, NY
- A. David Rothner, MD; Cleveland Clinic, Cleveland, OH

9:00 AM – 9:05 AM
CNS Bernard D’Souza International Fellowship Award

- Samson Gwer, MBChB, MRCPCH; Kenyatta University, Nairobi, Kenya

9:05 AM – 9:10 AM
Arnold P. Gold Foundation Humanism in Medicine Award at the Child Neurology Society:

- Douglas Postels, MD; Michigan State University, East Lansing, MI

9:15 AM – 12:00 PM
Symposium II: Presidential Symposium: Gene Therapy for Childhood Neurological Disease
Organizer: Mustafa Sahin, MD, PhD; Boston Children’s Hospital, Boston, MA
Introduction
Mustafa Sahin, MD, PhD

Gene therapy for muscular dystrophy: moving forward to new levels
Jerry R. Mendell, MD; Nationwide Children’s Hospital, Columbus, OH

Treatment of Ventilatory Failure in Pompe Disease by Gene Therapy: Next Generation Therapy
Barry Byrne, MD, PhD; University of Florida, Gainesville, FL

Developing Gene Delivery approaches for Spinal Muscular Arrophy and Translating to the Clinic
Brian K. Kaspar, PhD; Nationwide Children’s Hospital, Columbus, OH

Combination Therapy for Neurodegenerative Lysosomal Storage Diseases
Mark S. Sands, PhD; Washington University, St. Louis, MO

12:00 PM – 12:30 PM:
CNS Business Meeting

12:30 PM – 1:30 PM:
Learning in Child Neurology
Lawrence Faulkner, MD; President, American Board of Psychiatry and Neurology, Lake Forest, IL

1:45 PM – 4:00 PM:
Symposium III: Little Brains, Big Problems: Lasting Effects of Pediatric Traumatic Brain Injury
Organizer: Heidi Blume, MD, MPH; Seattle Children’s Hospital and Research Institute, Seattle, WA

Who Gives a Rat’s…Brain? Understanding the Translational Science of Pediatric TBI
Chris Giza, MD; Mattel Children’s Hospital, UCLA, Los Angeles, CA

Little Brains, Big Pains: Headache after Pediatric Concussion and TBI
Heidi Blume, MD, MPH

Waiting for the Other Shoe to Drop: Pediatric Post-traumatic Epilepsy
Howard Goodkin, MD, PhD; University of Virginia, Charlottesville, VA

Trajectories of Recovery and Predictors of Neurocognitive Outcome Following Pediatric TBI
Talin Babikian, PhD, ABPP; David Geffen School of Medicine at UCLA, Los Angeles, CA

Child Neuro News Break: Poster Walkaround/Wine & Cheese Reception (Non-CME)
Supported by Eisai, Inc.

7:00 – 8:15 AM:
Continental Breakfast and Seminars

Breakfast Seminar 4: Beyond “Seizure Disorders”: the New Classification of the Epilepsies
Organizers: Renée Shellhaas, MD and Sucheta Joshi, MD; University of Michigan, Ann Arbor, MI

The 2010 ILAE Proposed New Organization of the Epilepsies: What You Need to Know
Elaine Wirrell, MD; Mayo Clinic, Rochester, MN

What Happens When a Patient Doesn’t Fit in One of the New Boxes? Criticisms and Potential Difficulties with the New System
Peter Camfield, MD; Dalhousie University, Halifax, NS
How Do I Use this in my Practice?
What is “Done” and What is Up for Discussion? Practical Implementation in Clinical Practice and Research Proposals
Dennis Dlugos, MD, MSCE; Children's Hospital of Philadelphia, Philadelphia, PA

Breakfast Seminar 5: Next Generation Sequencing, Genomics, and Neurogenetics
Organizer: Tyler Mark Pierson, MD, PhD; Cedars-Sinai Medical Center, Los Angeles, CA
Next Generation Sequencing and Genomics: a Technological Introduction
David Adams, MD, PhD; NIH-Undiagnosed Diseases Program, Bethesda, MD
Neurogenetics and Genomics: Undiagnosed Cases in Pediatric Neurology
Tyler Mark Pierson, MD, PhD
Clinical Applications of Exome Sequencing: from the NICU to the Neurogenetics Clinic
Eric Marsh, MD, PhD; Children's Hospital of Philadelphia, Philadelphia, PA

Breakfast Seminar 6: Ethical Considerations in Gene Therapy
Organizer: Pedro Weisleder, MD, PhD; Nationwide Children's Hospital, Columbus, OH
Panel Discussion: “Gene Therapy: Proceed with Extreme Caution”
• Leon Epstein, MD; Ann & Robert Lurie Children's Hospital of Chicago, Chicago, IL
• William Graf, MD, PhD; Yale University School of Medicine, New Haven, CT
• Geoffrey Miller, MD, PhD; Yale University School of Medicine, New Haven, CT
• Pedro Weisleder, MD, PhD

8:30 AM – 10:15 AM
Platform Sessions 1 & 2

Platform Session 1:
8:30 AM – 8:45 AM
PS1-1 Darras BT et al
Results of a First-in-Human Phase I Study to Assess the Safety, Tolerability, and Dose Range Finding of a Single Intrathecal Dose of ISIS-SMNRC in Patients with Spinal Muscular Atrophy

8:45 AM – 9:00 AM
PS1-2 Raymond GV et al
Efficacy and Safety of Hematopoietic Cell Therapy in X-linked Adrenoleukodystrophy: a Multinstitutional Study (ALD-101)

9:00 AM – 9:15 AM
PS1-3 Karacy B et al
Viral Vector-based RNAi Gene Therapy for Alexander Disease

9:15 AM – 9:30 AM
PS1-4 Stasheff SF et al
Early Rpe65 Gene Therapy More Effectively Restores Precise Visual Responses to Retinal Ganglion Cells of Rpe65-/- Mice in vitro

9:30 AM – 9:45 AM
PS1-5 Schor NS et al
The p75 Neurotrophin Receptor and Autism

9:45 AM – 10:00 AM
PS1-6: Kaufmann WE et al
Randomized, Controlled, Phase2 Trial of STX209 (Arbaclofen) for Social Function in Autism Spectrum Disorder

10:00 AM – 10:15 AM
LATE BREAKING ABSTRACT:
PL1-7 Shirley MD et al
A Somatic Mosaic Mutation in GNAQ causes Sturge-Weber syndrome and Isolated Port-wine Birthmarks

8:30 AM – 8:45 AM
PS2-1 Tully HM
A New Approach to the Classification of Developmental Hydrocephalus

8:45 AM – 9:00 AM
PS2-2 Kuban KCK
Extremely Low Gestational Age Newborns (ELGANs) with Repeatedly Elevated Blood Concentrations of Inflammation-Related Proteins within 15 days of Birth are at Higher Risk of Cerebral Palsy

9:00 AM – 9:15 AM
PS2-3 Harbert MJ et al
Therapeutic Hypothermia is Associated with Improved Outcomes in Perinatal Stroke

9:15 AM – 9:30 AM
PS2-4 Schulze A
The Natural History of Late Infantile CLN2 Disease: Striking Homogeneity of Clinical Progression in Two Independently Obtained Large Clinical Cohorts

9:30 AM – 9:45 AM
PS2-5 Mallick AA et al
The Presenting Features of Arterial Ischaemic Stroke in a Population-Based Cohort

9:45 AM – 10:00 AM
PS2-6 McKnight D et al
A 53-gene Sequencing and Deletion/Duplication Panel Reveals a Broader Spectrum of Genotype-Phenotype Correlations in Epilepsy.

10:00 AM – 10:15 AM
LATE BREAKING ABSTRACT:
PS2-7 Anderson HM et al
Preventable Infections in Children with Leukodystrophies
10:45 AM – 10:55 AM
Child Neurology Society Awards

Outstanding Junior Member Awards

- Anuja Jindal, MD; Children's Hospital Pittsburgh, Pittsburgh, PA
- Archana Patel, MD; Children's Hospital Boston, Boston, MA
- Pilar Pichon, MD; Loma Linda University Medical Center, Loma Linda, CA
- Mark Schomer, MD; Children's Hospital Boston, Boston, MA
- Mitchel Williams, MD; Children's Hospital of Michigan, Detroit, MI

M. Richard Koenigsberger Scholarship Award

- Louis Dang, MD, University of Michigan, Ann Arbor, MI

10:55 – 11:00 AM
CNS/PCN Blue Bird Circle Training Program Director Award:

- Harvey Singer, MD; Johns Hopkins University, Baltimore, MD

11:00 AM – 11:10 AM
Child Neurology Foundation Scientific Awards Announcements

11:10 AM – 11:40 AM
Philip R. Dodge Young Investigator Award Lecture:

Peter T. Tsai, MD, PhD; Boston Children's Hospital, Boston, MA
Mechanisms Underlying the Cerebellar Contribution to Autism in Mouse Models of Tuberous Sclerosis

11:40 AM – 12:40 PM
Bernard Sachs Lecture:

Tallie Z. Baram, MD, PhD; University of California Irvine, Irvine, CA
Shaping the 4-Dimensional Brain

9:00 AM – 2:30 PM:
Exhibits

12:45 – 2:30 PM:
Lunch & Poster/Exhibit Walkaround

2:30 – 4:45 PM
Symposium IV: Treatable Genetic-Metabolic Epilepsies

Organizer: Phillip L. Pearl, MD; Children’s National Medical Center, Washington, DC

Treatable Genetic-Metabolic Epilepsies: Overview and Case Studies
Phillip L. Pearl, MD

Pyridoxine-Dependencies—Clinical and Research Update
Sidney M. Gospe, Jr., MD, PhD; Seattle Children's Hospital, Seattle, WA

Glucose Transporter 1 Deficiency—DeVivo Disease
Darryl C. DeVivo, MD; Columbia University, New York, NY

New Genes in Early Onset Epileptic Encephalopathies
Annapurna Poduri, MD; Boston Children’s Hospital, Boston, MA

4:45 – 6:00 PM:
Junior Member Seminar 1: Meet the Editors
Organizer: Renée Shellhaas, MD; University of Michigan, Ann Arbor, MI

Panel Members:

- John Bodensteiner, MD
- Amy Brooks-Kayal, MD, PhD
- Tobias Loddenkemper, MD, PhD
- E. Steve Roach, MD

Junior Member Seminar 2: Private Practice Child Neurology
Organizer: Jasna Kojic, MD; Orlando, FL
Discussant: Ashraf El-Bohy, MD; Orlando, FL

7:00 – 10:00 PM:
RECEPTION

Saturday, November 2

7:00 – 8:15 AM
CONTINENTAL BREAKFAST AND SEMINARS

Breakfast Seminar 7: Updates on Inherited Neuromuscular Disorders
Organizer: Peter Kang, MD; Boston Children’s Hospital, Boston, MA and University of Florida, Gainesville, FL

Duchenne Muscular Dystrophy: exon skipping strategies
Edward M. Kaye, MD; Sarepta Therapeutics, Cambridge, MA

Spinal Muscular Atrophy: emerging therapies
Basil T. Darras, MD; Boston Children’s Hospital, Boston, MA

Myotonic Dystrophy: genetic and therapeutic advances
Peter Kang, MD

Breakfast Seminar 8: Neuro-ophthalmology for the Child Neurologist: Practical clinical pearls
Organizer: Steven F. Stasheff, MD, PhD; University of Iowa, Iowa City, IA

Examining the Back of the Eye to Help Diagnose and Treat Pediatric Neuologic Disease
Steven F. Stasheff, MD, PhD
Updates on the Diagnosis of Papilledema, Pseudopapilledema, and IIH
Robert Avery, MA, DO; Children’s National Medical Center, Washington, DC

Congenital Ocular Motor Apraxia and Associated Neurodevelopmental Abnormalities
Michael S. Salman, MRCP, PhD; University of Manitoba, Winnipeg, MB

Breakfast Seminar 9: To Err is Human: Reducing Medical Errors by Better Handoffs with I-PASS
Organizer: James F. Bale, Jr., MD; University of Utah and Primary Children’s Hospital, Salt Lake City, UT
Panel Discussion:
- James F. Bale, Jr., MD
- David K. Urion, MD; Boston Children’s Hospital, Boston, MA
- Ann Yeh, MD; Hospital for Sick Children, Toronto, ON

9:00 – 9:50 AM  Hower Award Lecture: Genetic Bottlenecks and the Neurologic Diseases of the Navajo Native Americans
John Bodensteiner, MD; Mayo Clinic, Rochester, MN

10:00 AM – 12:15 PM  Symposium V: Paroxysmal Disorders
Organizer: Jonathan W. Mink, MD, PhD; University of Rochester Medical Center, Rochester, NY

Topics/ Speaker:
- Mechanistic Insights on Hypokalemic Periodic Paralysis Reveal a New Therapeutic Strategy
  Stephen C. Cannon, MD, PhD; University of Texas Southwestern, Dallas, TX
- Episodic Ataxias
  Abigail Collins, MD; Children’s Hospital Colorado, Denver, CO
- Alternating Hemiplegia of Childhood
  Kathryn Swoboda, MD; University School of Medicine, Salt Lake City, UT
- Paroxysmal Dyskinesias
  Jonathan W. Mink, MD, PhD
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<th>Time</th>
<th>Session</th>
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<tr>
<td>7:00 PM – 9:00 PM</td>
<td>ACNN Welcome Reception (Nurses only)</td>
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**Wednesday October 30, 2013**

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<th>Time</th>
<th>Session</th>
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<tr>
<td>7:00 AM – 8:00 AM</td>
<td>Registration and Continental Breakfast</td>
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<td>8:00 AM – 8:15 AM</td>
<td>Welcome and Introduction</td>
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<td>8:15 AM – 9:00 AM</td>
<td>Janet Brucker Keynote Address: TBA</td>
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<td>9:00 AM – 10:00 AM</td>
<td>Duchenne Muscular Dystrophy: Early Diagnosis and Care</td>
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<td>Kathi Kinnet, MSN, CNP</td>
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<td>10:00 AM – 10:15 AM</td>
<td>Break</td>
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<td>10:15 AM – 10:45 AM</td>
<td>Myasthenia Gravis: OMG X 3</td>
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<td>Jennifer Boyd RN, MHSc, CNN(C)</td>
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<td>10:45 AM – 11:30 AM</td>
<td>Stroke in Children: Now you see it, now you don’t</td>
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<td>Ivanna Yau RN, MN, NP-Peds, CNN (C)</td>
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<td>11:30 AM – 12:00 PM</td>
<td>Awards and Annual Business Meeting</td>
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<td>12:00 PM – 1:00 PM</td>
<td>Lunch</td>
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<td>1:00 PM – 2:00 PM</td>
<td>Is Intranasal or Buccal Midazolam as Effective as Rectal Diazepam in the Treatment of active Convulsive Seizures in the Pediatric Setting Outside the Hospital?</td>
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<td>Ann Morgan MS, RN, CPNP, CPN and Vicki Netzke-Doyle MSN, RN, CPN</td>
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**Thursday October 31, 2013**

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<tr>
<td>12:00 PM – 1:00 PM</td>
<td>Lunch-SIG</td>
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<td>Topics to be announced</td>
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<td>1:00 PM – 2:00 PM</td>
<td>A Randomized Controlled Trial of the Acceptability, Feasibility, and Preliminary Effects of Cognitive Behavioral Skills Building Intervention in Adolescents with Chronic Daily Headaches</td>
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<td>Carolyn Hickman, PhD, RN, CPNP</td>
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<td>2:45 PM – 3:00 PM</td>
<td>Improving Headaches Self-management Skills Using Medical Media in Children and Adolescents Age 8-18</td>
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<td>Ruth Rosenblum DNP, RN, PNP-BC</td>
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