

Advancing Hope: Pioneering Therapy for Rare Diseases



center for
rare disease
therapy

Friday, Feb. 19, 2016

8 a.m. to 6 p.m.

Children's Hospital of Pittsburgh of UPMC

**John G. Rangos Sr. Conference Center
Pittsburgh, PA**

*Sponsored by the Center for Rare Disease Therapy at
Children's Hospital of Pittsburgh of UPMC and the
University of Pittsburgh School of Medicine, Center
for Continuing Education in the Health Sciences*



GUEST SPEAKER

Senator Robert P. Casey, Jr.

Sponsor of the Advancing Hope Act of 2015, which extends and strengthens the U.S. Food and Drug Administration's priority review voucher program for rare, life-altering pediatric diseases, including all pediatric cancers and sickle cell disease. It also allows the program to continue indefinitely.



KEYNOTE SPEAKER

Edwin Naylor, PhD, MPH

Expert in newborn screening

*2014 Recipient of the
Robert Guthrie Award*



COURSE DIRECTOR

Jerry Vockley, MD, PhD

Chief, Medical Genetics

*Director, Center for Rare
Disease Therapy*

*Children's Hospital of Pittsburgh
of UPMC*

*Cleveland Family Professor
of Pediatric Research*

Professor of Human Genetics

*University of Pittsburgh
School of Medicine*



center for rare disease therapy

Join specialists from Children's Hospital of Pittsburgh of UPMC and a distinguished faculty of international experts for **Advancing Hope: Pioneering Therapy for Rare Diseases**. This inaugural one-day symposium will highlight pioneering treatments for rare conditions in children.

The symposium is free and available by live video stream at CHP.edu/AdvancingHope. CME credits are available for on-site participants only.

Who Should Attend

This symposium is designed for medical geneticists, neonatologists, gastroenterologists, pediatric surgeons, pediatricians, family practitioners, genetic counselors, nurses, mid-level providers, and parents/guardians who care for children with rare diseases. All other health professionals are also welcome.

Learning Objectives

- Physicians will be able to apply diagnostic algorithms and determine appropriate referral pathways for patients with rare diseases.
- Illustrate successful models for multidisciplinary collaborations that are essential for developing novel approaches to therapy for rare disease.
- Identify population-specific methods for genetic testing and disease treatment in the Plain populations and demonstrate successful application to other groups.

Continuing Education Credit

The University of Pittsburgh School of Medicine is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians.

The University of Pittsburgh School of Medicine designates this live activity for a maximum of **7.25 AMA PRA Category 1 Credits™**. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Other health care professionals are awarded **0.7** continuing education units (CEU's) which are equal to **7.2** contact hours.

Agenda

8 – 8:15 a.m.

Welcome

Jodie Vento, MGC, LCGC

Manager, Center for Rare Disease Therapy
Children's Hospital of Pittsburgh of UPMC

Michael Drnach, MBA

Executive Director, Children's Subspecialty Services
Children's Hospital of Pittsburgh of UPMC

8:15 – 8:45 a.m.

The Power of Translational Research

Jerry Vockley, MD, PhD

8:45 – 9:15 a.m.

Rare Disease Gene Discovery in Plain Populations

Erik Puffenberger, PhD

9:15 – 9:30 a.m.

Validating Candidate Variants of Interest: Principles into Practice

Karlla Brigatti, MS, LCGC

9:30 – 10 a.m.

Propionogenesis in Brain and Heart — Implications for Management Two European Disorders of Propionate Metabolism: PCCB.c1606A>G and Cbl-C Deficiency MMACHC c.271dupA.

D. Holmes Morton, MD

10 – 10:15 a.m.

Panel Questions

*Jerry Vockley, D. Holmes Morton, Erik Puffenberger,
Karlla Brigatti*

10:15 – 10:30 a.m.

Break

10:30 – 11:10 a.m.

Rare Genetic Disorders of the Eye and Novel Treatment Modalities

Ellen Mitchell, MD

11:15 – 12 p.m.

SPECIAL PRESENTATION: (Non-CME) The Advancing Hope Act

Senator Robert Casey, Jr.

Noon – 1 p.m.

Lunch

1:15 – 1:45 p.m.	Outcomes of Umbilical Cord Blood Transplantation for the Treatment of Leukodystrophies and Mucopolysaccharidoses <i>Maria Escolar, MD, MS</i>
1:45 – 2:15 p.m.	Reduced Intensity Conditioning for Bone Marrow/Stem Cell Transplant in Non-Malignant Disorders <i>Mark Vander Lugt, MD</i>
2:15 – 2:45 p.m.	Health Beliefs among Females Diagnosed and At-Risk for Fabry Disease <i>Katie Long, MS, LCGC</i>
2:45 – 3 p.m.	Panel Questions <i>Maria Escolar, Mark Vander Lugt, Katie Long</i>
3 – 3:15 p.m.	Break
3:15 – 3:45 p.m.	Neonatal Cholestasis: A Family of Rare Diseases <i>Robert Squires, MD</i>
3:45 – 4:15 p.m.	Total Pancreatectomy and Islet Cell Autotransplant to Treat Chronic Pancreatitis <i>Mark Lowe, MD, PhD</i>
4:15 – 4:45 p.m.	Pediatric Liver Transplantation for Metabolic Disease and the Role of Living Donor Transplantation <i>George Mazariegos, MD, FACS</i>
4:45 – 5 p.m.	Panel Questions <i>George Mazariegos, Mark Lowe, Robert Squires</i>
5 – 5:45 p.m.	CLOSING KEYNOTE: The Next Generation of Newborn Screening: Early Therapeutic Intervention <i>Edwin Naylor, PhD, MPH</i>
5:45 p.m.	Closing Remarks <i>Jerry Vockley, MD, PhD</i>
6 p.m.	Conference Adjournment

8:15– 8:45 a.m.



The Power of Translational Research

Jerry Vockley, MD, PhD

Chief, Medical Genetics

Director, Center for Rare Disease Therapy

Children's Hospital of Pittsburgh of UPMC

Cleveland Family Professor of Pediatric Research

Professor of Human Genetics

University of Pittsburgh School of Medicine

NOTES:

[illegible]

8:45 – 9:15 a.m.



**Rare Disease Gene Discovery
in Plain Populations**

Erik Puffenberger, PhD
Laboratory Director
Clinic for Special Children

NOTES:



Validating Candidate Variants of Interest: Principles into Practice

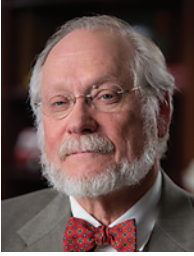
Karlla Brigatti, MS, LCGC

Genetic Counselor

Clinic for Special Children

NOTES:

[illegible]



Propionogenesis in Brain and Heart — Implications for Management Two European Disorders of Propionate Metabolism: PCCB.c1606A>G and Cbl-C Deficiency MMACHC c.271dupA.

D. Holmes Morton, MD

Co-Founder

Clinic for Special Children

NOTES:

This image shows a single sheet of white paper with horizontal blue or grey ruling lines. The lines are evenly spaced and run across the width of the page. There are approximately 20 lines visible. The paper has a slight shadow on the right side, suggesting it's resting on a surface.

11:15 a.m. – 12 p.m.



SPECIAL PRESENTATION: (Non-CME)
The Advancing Hope Act

Senator Robert Casey, Jr.

NOTES:



Outcomes of Umbilical Cord Blood Transplantation for the Treatment of Leukodystrophies and Mucopolysaccharidoses

Maria Escolar, MD, MS

Director, Program for the Study of Neurodevelopment
in Rare Disorders

Children's Hospital of Pittsburgh of UPMC

Associate Professor of Pediatrics

University of Pittsburgh School of Medicine

NOTES:

This image shows a single sheet of white paper with horizontal ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.

1:45 – 2:15 p.m.



Reduced Intensity Conditioning for Bone Marrow/Stem Cell Transplant in Non-Malignant Disorders

Mark Vander Lugt, MD

Pediatric Hematology/Oncology

Children's Hospital of Pittsburgh of UPMC

NOTES:

[illegible]



Health Beliefs among Females Diagnosed and At-Risk for Fabry Disease

Katie Long, MS, LCGC

Genetic Counselor

Children's Hospital of Pittsburgh of UPMC

NOTES:

This image shows a single sheet of white paper with horizontal blue ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.



Neonatal Cholestasis: A Family of Rare Diseases

Robert Squires, MD

Director, Pediatric Hepatology Program
Children's Hospital of Pittsburgh of UPMC
Professor of Pediatrics
University of Pittsburgh School of Medicine

NOTES:

This image shows a blank sheet of white paper with horizontal ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.

3:45 – 4:15 p.m.



Total Pancreatectomy and Islet Cell Autotransplant to Treat Chronic Pancreatitis

Mark Lowe, MD, PhD

Chief, Pediatric Gastroenterology, Hepatology and Nutrition
Children's Hospital of Pittsburgh of UPMC
Carol Ann Craumer Endowed Chair for Pediatric Research
Professor and Vice-Chairman of Pediatrics
University of Pittsburgh School of Medicine

NOTES:



Pediatric Liver Transplantation for Metabolic Disease and the Role of Living Donor Transplantation

George Mazariegos, MD, FACS

Hillman Center for Pediatric Transplantation

Children's Hospital of Pittsburgh of UPMC

Jamie Lee Curtis Professor of Surgery and

Critical Care Medicine

University of Pittsburgh School of Medicine

NOTES:

This image shows a blank sheet of white paper with horizontal ruling lines. The lines are evenly spaced and extend across the width of the page. There are no margins, text, or other markings on the paper.



CLOSING KEYNOTE:

The Next Generation of Newborn Screening: Early Therapeutic Intervention

Edwin Naylor, PhD, MPH

Expert in newborn screening

2014 Recipient of the Robert Guthrie Award

NOTES:

This image shows a single sheet of white paper with horizontal blue or grey ruling lines. The lines are evenly spaced and run across the width of the page. There are approximately 20 lines visible. The paper has a slight shadow on the right side, suggesting it's resting on a surface.

Faculty Disclosure

All individuals in a position to control the content of this education activity are required to disclose all relevant financial relationships with any proprietary entity producing, marketing, re-selling, or distributing health care goods or services, used on, or consumed by, patients.

No relevant financial relationships with commercial entities were disclosed by:

Karla Brigatti, MS, LCGC

Michael Drnach, MBA

Maria Escolar, MD, MS

Mark Vander Lugt, MD

George Mazariegos, MD, FACS

Ellen Mitchell, MD

Erik Puffenberger, PhD

Robert Squires, MD

Jodie Vento, MGC, LCGC

The following information was disclosed:

Katie Long, MS, LCGC receives grant research support from Sanofi Genzyme.

Mark Lowe, MD, PhD is a consultant for Nordmark Arzneimittel GmbH & Co. and he receives royalties from Millipore, Inc.

D. Holmes Morton, MD is a consultant with Cambrooke Therapeutics and unpaid research collaborator with Agios Pharmaceuticals.

Edwin Naylor, PhD, MPH is a consultant for Parabase Genomics.

Jerry Vockley, MD, PhD is a consultant for AGT and receives grant research from Ultragenyx, Mitobridge, and Horizon Pharma.

The information presented at this CME program represents the views and opinions of the individual presenters, and does not constitute the opinion or endorsement of, or promotion by, the UPMC Center for Continuing Education in the Health Sciences, UPMC / University of Pittsburgh Medical Center or Affiliates and University of Pittsburgh School of Medicine. Reasonable efforts have been taken intending for educational subject matter to be presented in a balanced, unbiased fashion and in compliance with regulatory requirements. However, each program attendee must always use his/her own personal and professional judgment when considering further application of this information, particularly as it may relate to patient diagnostic or treatment decisions including, without limitation, FDA-approved uses and any off-label uses. The Center for Continuing Education and the sponsoring department do not certify that through participation in this program participants are competent to perform certain procedures or skills.

The University of Pittsburgh is an affirmative action, equal opportunity institution.

Center for Rare Disease Therapy



We are international experts focused on treating children with rare diseases, defined by pioneering protocols and individualized services in a world-class environment.

www.chp.edu/rarecare

Contact Us

For an appointment or consultation, please contact **Jodie Vento, MGC, LCGC**, at 412-692-RARE (7273) or RareCare@chp.edu.

center for
rare disease
therapy



Children's | of
Hospital of Pittsburgh | UPMC

4401 Penn Ave. | Pittsburgh, PA 15224 USA



4401 Penn Ave.
Pittsburgh, PA 15224

www.chp.edu