



OLIVIA'S LIGHT
Pediatric Rare Disease
Conference

Keeping Genetics Top of Mind

June 9-10, 2025

UVA Student Health & Wellness
550 Brandon Ave, Charlottesville, VA



Program Overview

The Olivia's Light Pediatric Rare Disease Conference aims to improve patient care for children and families impacted by a rare disease. It will highlight aspects of the rare disease journey, with an emphasis on rapid broad genetic testing, ethical considerations, and strategies to empower families.

Target Audience

All stakeholders in pediatric rare diseases, including but not limited to: Pediatric Geneticists, Developmental specialists, Pediatric Neurologists, Neonatologists and other Pediatric subspecialties, Maternal Fetal Medicine, Physician Associates, Nurse Practitioners, Nurses, Genetic Counselors, Clinical Psychologists, Pediatricians, Dietitians, Physical Therapist, Occupational Therapist, Respiratory Therapist, Speech Pathologists, Social Workers

Families and others involved in this area are welcome to attend.

Educational Outcomes

1. Describe the diagnostic odyssey for families of children with undiagnosed genetic conditions.
2. Describe the evidence for the need of rapid genetic testing in acute setting.
3. Identify the impact of rapid testing on patient care: where both positive and negative results matter.
4. Identify a novel pathway to access whole exome sequencing
5. Identify key prenatal phenotypes and neonatal presentations that may indicate underlying genetic disorders.
6. Apply comprehensive imaging techniques to characterize phenotypes and achieve early diagnosis of genetic conditions.
7. Implement shared decision-making from multiple lenses: contrasting parent and provider perspectives
8. Define ethical considerations related to prenatal/neonatal consultations
9. Describe the development of a novel clinical telehealth model providing behavioral interventions and caregiver training for individuals with fragile X syndrome (FXS)

Course Directors

Jenna King

Olivia's Light Co-Founder & President
Parent and Patient Advocate

David Kaufman, MD, FAAP

Neonatal Perinatal Medicine
University of Virginia Health Children's

Invited Guest Faculty

Ricki S Carroll, MD, MBE

Skeletal Dysplasia and Palliative Care
Pediatrician
Nemours Children's Hospital

Paul Kruszka, MD

Chief Medical Officer
GeneDx

Bimal P Chaudhari, MD, MPH

Assistant Professor, Pediatrics –
Neonatal Genetics
Nationwide Children's Hospital

Dmitriy Niyazov, MD

Associate Professor of Pediatrics,
Medical Genetics
Duke University School of Medicine

Michelle Curtin, DO

Associate Professor, Pediatrics -
Developmental
Wake Forest University School of
Medicine

Debra Reisinger, PhD

Pediatric Psychologist, Behavioral
Medicine
Cincinnati Children's Hospital

Jamie Fraser, MD, PhD

Director of Myelin Disorders and
Prenatal Genetics Programs
Children's National Hospital

Amanda Soroka, NP*

Pediatric Nurse Practitioner, Fetal
Center
Children's Hospital of The Kings
Daughters

Jennifer Harmon, MD, PhD

Assistant Professor, Pediatric
Neurology, Genetics
Wake Forest University School of
Medicine

Connie Williams, MD, PhD

Associate Professor, Pediatrics
McMaster University, Ontario, Canada

Elizabeth Jalazo, MD

Assistant Professor, Pediatrics
University of North Carolina School of
Medicine

UVA Faculty

Paula Darradji, RN

RN Case Manager

Joy Miller, PNP, CHPPN

APP Manager, Pediatric Critical Care

Julia Figueroa, RN, BSN*

Home Health, UVA Health Continuum

Anna Platt, MS, LCGC*

Pediatric Genetic Counseling

Sarah M Lepore, MSN, APRN, NNP-BC

APP Neonatal ICU Manager

Lisa Zuckerwise, MD

Director, Maternal-Fetal Medicine
Program

Additional Planning Committee

Melanie Rasnic, PA

Pediatric Genetics
Virginia Commonwealth University
Health

Christina Peroutka, MD

Assistant Professor, Pediatrics -
Genetics
UVA Health Children's

Haley Life, RN

Newborn ICU
UVA Health Children's

Kara Watts

Program Manager, Continuing
Education
UVA Health

Katheryn Frazier, MD

Associate Professor, Pediatrics –
Developmental
UVA Health Children's

* indicates Planning Committee

Monday, June 9, 2025

7:00AM – 7:50AM

Registration, Breakfast (provided), and Exhibits

8:00AM – 8:30AM

Welcome, Conference Overview & Goals, and Parent Testimony (CME)

Jenna King, Parent

8:30AM – 9:30AM

KEYNOTE: Exceptional Care without Exception: Ending Genetic Exceptionalism to Advance Child Health (CME)

Bimal P Chaudhari, MD, MPH

9:30AM – 10:30AM

Rapid Exome & Genome: Fast Diagnosis is Necessary to Improve Patient Care (CME)

Dmitriy Niyazov, MD

10:30AM – 11:00AM

Network Break/Exhibits

11:00AM – 11:20AM

Parent Testimony (non-CME)

Amanda Soroka, NP

11:20AM – 12:20PM

Shortening the Diagnostic Odyssey: Current and Genomic Testing on the Horizon (non-CME)

Paul Kruszka, MD

12:20PM – 12:45PM

The Expanding Role of Genetic Counselors and What it Means for Patients (non-CME)

Anna Platt, MS, LCGC

12:45PM – 1:45PM

Lunch (provided) & Exhibits

1:50PM – 2:50PM

A Novel Pathway to Increase Whole Exome Sequencing Access for Pediatric Patients with Neurodevelopmental Disabilities (CME)

Michelle Curtin, DO

Jennifer Harmon, MD, PhD

2:50PM – 3:50PM

From Clinical Care to Community Outreach: A Framework for Increasing Access to Care in Individuals with Fragile X Syndrome and Other Rare Genetic Disorders (CME)

Debra Reisinger, PhD

3:50PM – 4:50PM

Early Recognition of Genetic Conditions: Prenatal and Neonatal Red Flags (CME)

Jamie Fraser, MD

4:50PM – 6:00PM

Networking Reception

Tuesday, June 10, 2025

7:00AM – 7:45AM

Registration, Breakfast (provided), and Exhibits

7:45AM – 8:00AM

Conference Overview & Today's Goals (CME)

Jenna King, Parent

David Kaufman, MD

8:00AM – 8:45AM

PANEL: The Rare Disease Journey from the Diagnosis and Beyond: Insights from Veteran Nurses in the Trenches (CME)

Paula Darradji, Nurse

Julia Figueroa, RN, BSN

Joy Miller, PNP, CHPPN

Sarah M Lepore, NP (Moderator)

8:45AM – 9:45AM

Small Populations, Big Breakthroughs: The Evolving Therapeutic Landscape for Rare Disease Patients (CME)

Elizabeth Jalazo, MD

9:45AM – 10:15AM

Network Break/Exhibits

10:15AM – 11:15AM

Facilitating Shared Decision Making in the Context of Rare Diagnoses and Uncertain Prognoses (CME)

Connie Williams, MD, PhD

11:15AM – 12:15PM

Teams, Technology and Families Changing Outcomes in Rare Diseases that were Formerly Considered "Lethal": Lessons Learned from Skeletal Dysplasia Patients (CME)

Ricki S Carroll, MD, MBE

12:15PM – 1:30PM

PANEL: The Weight of Our Words: Considerations in Prenatal/Neonatal Consultation (CME)

Bimal P Chaudhari, MD, MPH

Jamie Fraser, MD

Connie Williams, MD, PhD (Moderator)

Lisa Zuckerwise, MD

Ricki S Carroll, MD, MBE

1:30PM – 1:35PM

Adjourn with lunch provided to go

Accreditation and Designation Statements



In support of improving patient care, this activity has been planned and implemented by the UVA Health Continuing Education and Olivia's Light. UVA Health Continuing Education is jointly accredited by the Accreditation Council for Continuing Medical Education (ACCME), the Accreditation Council for Pharmacy Education (ACPE), and the American Nurses Credentialing Center (ANCC), to provide continuing education for the healthcare team.

This activity was planned by and for the healthcare team, and learners will receive 10.5 Interprofessional Continuing Education (IPCE) credit for learning and change.



Physicians

UVA Health Continuing Education designates this live activity for a maximum of 10.5 AMA PRA Category 1 Credits™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Nurses

UVA Health Continuing Education awards up to 10.5 contact hour(s) for nurses who participate in this educational activity and complete the post activity evaluation.

Hours of Participation

UVA Health Continuing Education awards 10.5 hours of participation (consistent with the designated number of AMA PRA Category 1 Credit(s)™ or ANCC contact hours) to a participant who successfully completes this educational activity. UVA Health Continuing Education maintains a record of participation for six (6) years.



Physician Associates

UVA Health Continuing Education has been authorized by the American Academy of PAs (AAPA) to award AAPA Category 1 CME credit for activities planned in accordance with AAPA CME Criteria.

This activity is designated for 10.5 AAPA Category 1 CME credits. PAs should only claim credit commensurate with the extent of their participation.

Registration and Fees

Registration and payment are available at the UVA Continuing Medical Education portal www.cmevillage.com or the QR Code.

Registration will close midnight on Tuesday, May 27, 2025. **No walk in registration.** Syllabus will be available online the week of the conference until 30 days after the program. See cancellation policy prior to registration.



Register by May 27, 2025

Physician or PhD	\$275
Other HCP/Trainee	\$140
Parents or Advocates	\$99
Industry or Government	\$275

Contact Information

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