

MECP2 Duplication Syndrome Conference
Jan and Dan Duncan Neurological Research Institute
September 23 – September 24, 2022

FRIDAY, SEPTEMBER 23

- 8:15 – 8:55 **Registration and Continental Breakfast**
Jan and Dan Duncan Neurological Research Institute
7th Floor Conference Center
- 9:00 – 9:15 **Huda Y. Zoghbi, MD**
Welcome Remarks
Howard Hughes Medical Institute
Baylor College of Medicine
Jan and Dan Duncan Neurological Research Institute
- 9:15 – 9:45 **Zhenya Ivakine, PhD**
**Novel Mecp2-Irak1 duplication mouse model: implications for the
CRISPR/Cas9-based genome editing in MECP2 duplication syndrome**
Department of Physiology, The Hospital for Sick Kids
Toronto, Canada
- 9:45 – 10:15 **Hilde Van Esch, MD, PhD**
**Study of human MDS neurons to better understand MECP2 duplication
syndrome**
Department of Human Genetics
KU Leuven, Belgium
- 10:15 – 10:35 **COFFEE BREAK**
- 10:35 – 11:00 **Sarika Peter, PhD**
Development of Clinician-reported Outcomes in MECP2 Duplication Syndrome
Vanderbilt University Medical Center
- 11:00 – 11:30 **Sameer Bajjkar, PhD / Ashley Grace-Anderson, PhD**
Update on studies and regulation of MeCP2 levels
Baylor College of Medicine
- 11:30 – 12:00 **Jeffrey L. Neul, MD, PhD**
Development of Severity Scale in MECP2 Duplication Syndrome
Vanderbilt University Medical Center
- 12:00 – 1:15 **LUNCH BREAK**
- 1:15 – 1:45 **Frank Rigo, PhD / Paymaan Jafar-Nejad, MD**
Efficacy, Tolerability and Toxicity Studies with ASO treatment
Ionis Pharmaceuticals

- 1:45 – 2:10 **Mirjana Maletic-Savatic, MD, PhD**
Biomarker Discovery in MECP2 Duplication Syndrome
Baylor College of Medicine
- 2:10 – 2:30 **COFFEE BREAK**
- 2:30 – 2:55 **Eric Marsh, MD, PhD**
Epilepsy in MECP2 Duplication Syndrome
Children’s Hospital of Philadelphia
- 2:55 – 3:20 **Kathleen Motil, MD, PhD**
Gastrointestinal Health in MECP2 Duplication Syndrome
Baylor College of Medicine
- 3:20 – 3:55 **Holly Harris, MD**
Developmental problems in MECP2 Duplication Syndrome
Baylor College of Medicine
- 3:55 – 4:25 **Christine Coquery, PhD**
**Establishing Natural History Data to Support the Drug Development Process
for MECP2 Duplication Syndrome**
Ionis Pharmaceuticals
- 4:25 – 5:00 **Family member time for open discussion**
- 5:30 – 6:30 **RECEPTION**
- 6:30 – 8:00 **DINNER**

SATURDAY, SEPTEMBER 24

- 9:00 – 9:25 **Davut Pehlivan, MD**
Genetics Studies in MECP2 Duplication Syndrome and its Impact on Treatment
Texas Children’s Hospital
- 9:25 – 9:50 **Marianna Sockrider, MD**
Pulmonary Care in MECP2 Duplication Syndrome
Texas Children’s Hospital
- 9:50 – 10:15 **Rochelle Dy, MD**
Rehabilitation Considerations in MECP2 Duplication Syndrome
Texas Children’s Hospital
- 10:15 – 10:40 **Danielle Edwards**
**The MECP2 duplication syndrome patient community views on living with
MDS**
Ionis Pharmaceuticals

10:40 – 11:00	COFFEE BREAK
11:00 – 11:25	Ivan K. Chinn, MD Frequent Infections in MECP2 Duplication Syndrome Texas Children's Hospital
11:25 – 11:50	Sherry Vinson, MD Burden on MDS Caregivers and Practical Tips for Parents Texas Children's Hospital
11:50 – 12:30	Bernhard Suter, MD and Huda Zoghbi, MD Wrap Up and Future Outlook
12:30 – 1:30	LUNCH (TO GO)
1:30 – 5:00	SOCIAL ACTIVITY FOR FAMILIES and CHILDREN