

# HNRNP-Related Neurodevelopmental Disorders 2023 Family and Scientific Symposium



**May 4th-May 6th, 2023**

New York, NY, USA



**COLUMBIA**

COLUMBIA UNIVERSITY  
IRVING MEDICAL CENTER

# Location



COLUMBIA

SCHOOL OF  
NURSING

560 W 168th St, New York, NY, 10032



# Speakers



**Maddie Gillentine, PhD**

Geneticist  
VP of HNRNP Family Foundation,  
Chair of Scientific and Medical  
Advisory Board,  
Symposium host,  
Variant Scientist,  
Seattle Children's Hospital



**Leila Margolis**

Founder and President of  
the HNRNP Family  
Foundation,  
Mom to Sidney who has  
SYNCRIP/HNRNPQ-RNDD,  
Symposium host



**Jennifer Bain, MD, PhD**

Scientific and Medical  
Advisory Board Member,  
Symposium host,  
Assistant Professor of  
Neurology and Pediatrics,  
Columbia University Irving  
Medical Center



**Christopher Ricupero, PhD**

Scientific and Medical Advisory  
Board Member,  
Symposium host,  
Associate Research Scientist,  
Columbia University Irving  
Medical Center



**Meena Balasubramanian  
MBBS, DCH, FRCPCH, MD**

Scientific and Medical Advisory  
Board Member,  
Symposium host,  
Senior Clinical Lecturer in  
Musculoskeletal Genetics,  
University of Sheffield



**Billie Au, MD**

Scientific and Medical  
Advisory Board Member,  
Symposium host,  
Clinical Assistant Professor of  
Medical Genetics and  
Pediatrics,  
University of Calgary



**Antonie Kline, MD**

Symposium host, Medical Director of Cleft Lip and Palate, Director of Clinical Genetics  
GBMC Healthcare



**Sean Post, PhD**

Associate Professor  
Department of Leukemia  
at MD Anderson Cancer Center



**Isabelle Draper, PhD**

Assistant Professor  
Molecular Cardiology  
Research Institute and  
Molecular Pharmacology  
Research Center, Tufts  
University



**Jane Juusola, PhD, FACMG**

Vice President  
GeneDx



**Barbara Corneo, PhD**

Director, Stem Cell Core,  
Columbia Stem Cell Initiative



**Prof. Stuart A. Wilson**

School of Biosciences  
University of Sheffield



**Cassidy Da Silva**

Doctoral Student  
University of Calgary



**Jo Ristow, MS, CCC-SLP**

Speech Language Pathologist  
CHAT NW



**Kalayla Hough, MS, CCC-SLP**

Speech Language Pathologist  
Magnolia Pediatric  
Therapy Collective

# Additional Colleagues and Supporters



**Kara Murias, MD,  
PhD, FRCPC**  
Assistant Professor  
of Pediatrics, Clinical  
Neurosciences,  
Psychiatry,  
Pediatric Neurologist  
University of Calgary



**Lindsay Phillips, PhD**  
Research Assistant  
Dept of Medical Genetics,  
University of Calgary



**Nicolò Pini, PhD**  
Department of Psychiatry,  
Columbia University  
Irving Medical Center and  
Division of Developmental  
Neuroscience, New York  
State Psychiatric Institute



**Marcia Ferguson, MGC**  
Genetic Counselor  
GBMC Healthcare



**Joanna Feng**  
Clinical Research  
Coordinator  
Columbia University  
Irving Medical Center



**Sonali Dalal Talsania, MD**  
Pediatric Ophthalmology  
Columbia University  
Irving Medical Center



**Aliaa Hamed Abdelhakim, MD, PhD**  
Pediatric Ophthalmology  
Columbia University  
Irving Medical Center



**Rachel Salazar**  
Physical Therapist  
Columbia University  
Irving Medical Center



**Manroop Chawla**  
Physical Therapist  
Columbia University  
Irving Medical Center



**Juliana Nitis**  
Medical Student  
Volunteer Coordinator



**Nicole Holuba**  
Pediatric Nurse Practitioner



**Thursday, May 4th**

**9:00AM-2:00PM**

8:45AM Coffee and Light Breakfast Hosted by the  
Columbia Tissue Engineering Resource Center

9:00AM

**Welcome!**

Maddie Gillentine, PhD and Leila Margolis  
*The HNRNP Family Foundation*

9:10AM

**An overview of the HNRNP-RNDDs**

Maddie Gillentine, PhD  
*The HNRNP Family Foundation,  
Seattle Children's Hospital*

**HNRNPU-Related Neurodevelopmental Disorder**

9:25AM

**HNRNPU-Related Disorder: What Do We Know So Far?**

Meena Balasubramanian, MBBS, DCH, FRCPCH, MD  
*The HNRNP Family Foundation,  
Sheffield Children's Hospital*

9:50AM

**The role of the hnRNPU gene family in transcription and  
splicing**

Prof. Stuart Wilson, PhD  
*School of Biosciences, University of Sheffield*

**HNRNPH2-Related Neurodevelopmental Disorder**

10:10AM

**HNRNPH2-RNDD and Simons Searchlight**

Jennifer Bain, MD, PhD  
*The HNRNP Family Foundation,  
Columbia University Irving Medical Center*

**Thursday, May 4th**  
**9:00AM-2:00PM**

10:35AM	<p><b>Exploring the HNRNP-RNDDs with iPSCs</b> Christopher Ricupero, PhD <i>The HNRNP Family Foundation, Columbia University Irving Medical Center</i></p>
<p><b>Break</b></p>	
<p><b>Au-Kline Syndrome (<i>HNRNPK</i>)</b></p>	
11:10AM	<p><b>Au-Kline Syndrome (<i>HNRNPK</i>)</b> Antonie Kline, MD <i>GBMC Healthcare</i></p>
11:35AM	<p><b>Episignatures of the HNRNP-RNDDs: Focus on <i>HNRNPK</i></b> Billie Au, MD <i>HNRNP Family Foundation University of Calgary</i></p>
12:00PM	<p><b>hnRNPK, a tightrope between development and tumorigenesis</b> Sean Post, PhD <i>MD Anderson</i></p>
<p><b>Working Lunch provided by ThermoFisher</b></p>	



**Thursday, May 4th**  
**9:00AM-2:00PM**

**Emerging HNRNP-Related Neurodevelopmental Disorders**

12:25PM

**HNRNPL-RNDD**  
Isabelle Draper, PhD  
*Tufts University*

12:50PM

**Teamwork makes the dream work: GeneMatcher and collaborations**  
Jane Juusola, PhD, FACMG  
*GeneDx*

1:10PM

**Closing Remarks: Whats next for the HNRNP-RNDDs?**

1:20PM

**Group Photo!**







1:30PM



**Networking**

**HNRNP-RNDD Patient Assessments 1:30PM-5:30PM**  
**Schedules to be distributed to families**

**Happy Hour @ 6pm**  
**Fort Washington Public House**  
**3938 Broadway, New York, NY 10032**



# Friday May 5th: Assessments

Test	Purpose	Clinician(s)/Researcher(s)
Medical Evaluation 	Determine clinical features of HNRNP-RNDDs	Dr. Meena Balasubramanian, Dr. Billie Au, Dr. Jennifer Bain, Dr. Antonie Kline, Marcia Ferguson, GC, Kara Murias, MD
Physical Examination 		Dr. Meena Balasubramanian, Dr. Billie Au, Dr. Jennifer Bain, Dr. Antonie Kline  3D Facial Imaging: Cassidy da Silva
Neurological Examination 		Dr. Jennifer Bain and Dr. Kara Murias
Motor Assessment 		Dr. Manroop Chawla and Dr. Rachel Salazar
Ophthalmology 		Dr. Sonali Talsania and Dr. Aliaa Abdelhakim
EEG 		Dr. Nicolò Pini, Jessica Hu, and Celia D'Amato

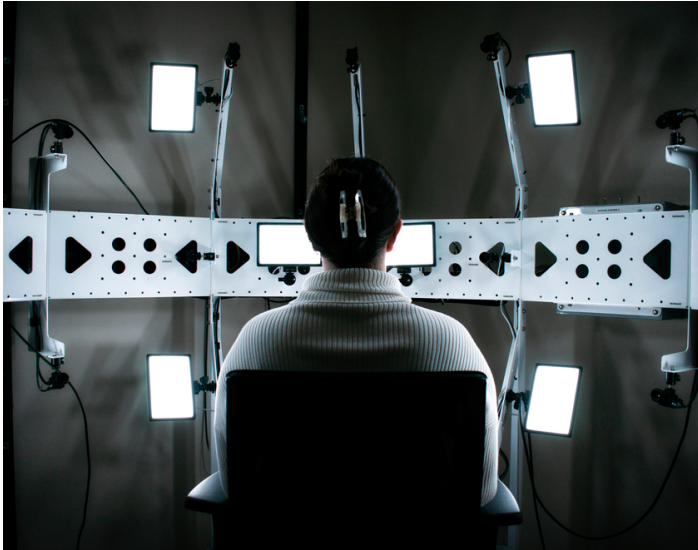
<p>Blood sample</p> 	<p>For methylation analysis, to generate induced pluripotent stem cells, for biomarker analysis</p>	<p>Simons Searchlight, Dr. Christopher Ricupero</p>
<p>Skin Biopsy</p> 	<p>To generate induced pluripotent stem cells</p>	<p>Dr. Meena Balasubramanian, Dr. Jennifer Bain, Dr. Christopher Ricupero, and Dr. Barbara Corneo</p>

## Details

Schedules to be distributed to families




<p>Medical Evaluation</p> 	<p>This is a meeting with one of our clinicians to go over medical history and assess medical needs. This should take 20-30 minutes. Families will receive a link prior to the meeting to fill in some medical history.</p>
<p>Physical Evaluation</p> 	<p>This is a meeting with one of our clinicians who is also a dysmorphologist (they really know what people look like!). This will take 15-20 minutes.</p>




## 3D Facial Imaging with Cassidy da Silva



Cassidy is a graduate student working with the investigators for the FaceBase study. This is an international study aiming to better characterize genetic syndromes that include facial effects and develop a computer-based tool to use facial imaging to help physicians diagnose these syndromes. Cassidy will take three-dimensional (3D) facial photographs of people and potentially their relatives.

This should take about 10-15 minutes.

<p>Neurological Examination</p> 	<p>This is a meeting with one of our clinicians to go assess neurological traits with tests like the Vineland Adaptive Behavior Scale. This should take 20-30 minutes. Tests may be sent to families to complete online.</p>
<p>Motor Assessment</p> 	<p>This is a meeting with one of our physical therapists. They will use tests to measure motor function. Videos may be taken to assess gait differences. This will take 20-30 minutes.</p>
<p>Ophthalmology</p> 	<p>This is a meeting with one of our pediatric ophthalmologists. They will assess and differences in vision and eye structure. This will take 20-30 minutes. Only families with <i>HNRNPK</i> variants/Au-Kline syndrome will undergo an ophthalmology assessment.</p>

<p style="text-align: center;">EEG</p> 	<p style="text-align: center;">This is an assessment for an electroencephalogram (EEG). The EEG cap to be used contain soft, noninvasive sensors that will be placed along the scalp. These will measure differences in brain activity. This will take about 30 minutes. Note if your child has had an EEG, this is different and collects more information.</p>
<p style="text-align: center;">Blood sample</p> 	<p style="text-align: center;">Simons Searchlight will be drawing blood at the family symposium. One sample of blood [up to 40ml, or less than 3 tablespoons] will be collected per participant. With the skin biopsy, this will take about 20 minutes.</p>
<p style="text-align: center;">Skin Biopsy</p> 	<p style="text-align: center;">A skin tissue sample [3mm] will be collected per participant (who consent). A punch biopsy technique will be used to obtain the skin sample. All samples will be stored in medium containing growth factors to keep the skin cells alive. The skin biopsies will be made into skin cells (fibroblasts) and induced pluripotent stem cells (iPSCs) and will be distributed to researchers after the meeting.</p> <p style="text-align: center;">With the blood draw, this should take about 20 minutes.</p>

**Lunch from 12:00PM-1:00PM**

**Hosted by the Weinberg Cerebral Palsy Center @  
Columbia University Irving Medical Center**

**Saturday, May 6th**  
**Morning Session: 10:00AM-12:30PM**

Light Breakfast 9:30AM	
10:00AM	<b>Welcome!</b> Maddie Gillentine, PhD and Leila Margolis <i>The HNRNP Family Foundation</i>
10:20AM	<b>A clinical overview of the HNRNP-RNDDs</b> Antonie Kline, MD <i>GBMC Healthcare</i>
10:45AM	<b>What is Simons Searchlight?</b> Erica Jones, MA <i>Simons Foundation</i>
Break	
Specific HNRNP-Related Neurodevelopmental Disorders	
11:15AM	<b>HNRNPH2-RNDD and our NHS</b> Jennifer Bain, MD, PhD <i>The HNRNP Family Foundation, Columbia University Irving Medical Center</i>
11:40AM	<b>HNRNPU-Related Disorder and our NHS</b> Meena Balasubramanian, MBBS, DCH, FRCPCH, MD <i>The HNRNP Family Foundation, Sheffield Children's Hospital</i>

**Saturday, May 6th**  
**Afternoon Session: 1:30PM-5:00PM**

12:05PM	<b>Au-Kline Syndrome (<i>HNRNPK</i>)</b> Antonie Kline, MD <i>GMBC Healthcare</i>
<b>Lunch Break from 12:30PM-1:30PM</b>	
<b>Functional Studies Focused on the <i>HNRNP</i> Genes</b>	
1:30PM	<b>Exploring the HNRNP-RNDDs with iPSCs</b> Christopher Ricupero, PhD <i>The HNRNP Family Foundation,</i> <i>Columbia University Irving Medical Center</i>
1:55PM	<b>The Columbia Stem Cell Core: helping labs by creating tools for disease modeling &amp; drug screening</b> Barbara Corneo, PhD <i>Columbia Stem Cell Initiative</i>
2:05PM	<b>Methylation Patterns of the HNRNP-RNDDs</b> Billie Au, MD <i>The HNRNP Family Foundation,</i> <i>University of Calgary</i>
2:30PM	<b>Developing 3D facial imaging data and tools to transform syndrome diagnosis</b> Cassidy Da Silva <i>University of Calgary</i>

# Saturday, May 6th

## Afternoon Session: 1:30PM-5:00PM

2:45PM	Group Photo!
<b>Break</b>	
<b>Learning from Each Other</b>	
3:20PM	<b>Understanding your child's genetic test result report</b> Jane Juusola, PhD, FACMG <i>GeneDx</i>
3:40PM	<b>Speech &amp; Language Therapy Considerations for Children Learning to Speak &amp; Communicate</b> Jo Ristow, MS, CCC-SLP and Kalayla Hough, MS, CCC-SLP <i>CHAT NW and Magnolia Pediatric Therapy Collective</i>
4:05PM	<b>Round Tables</b> Therapies: What works? Seizures Advocacy: How to engage clinicians in rare disorders?
4:30PM	<b>Town Hall</b> Ask the experts
5:00PM	Wrap Up

Saturday schedules for assessments to be distributed to families



We have a lot of people to thank for helping with our symposium!

## Sponsors



Weinberg Family  
Cerebral Palsy Center  
Columbia University Medical Center



## Travel Grants



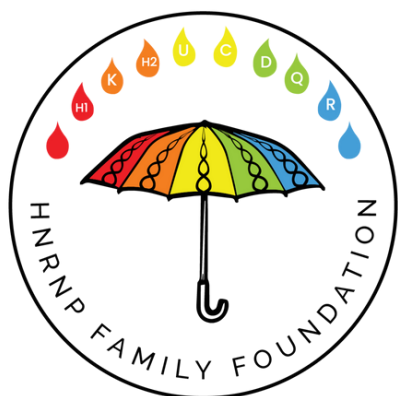
## Hosts

### Board

Leila Margolis  
Dr. Maddie Gillentine  
Dr. Susan Altschuller

### Scientific and Medical Advisory Board

Dr. Maddie Gillentine  
Dr. Jennifer Bain  
Dr. Christopher Ricupero  
Dr. Meena Balasubramanian  
Dr. Billie Au  
Dr. Antonie Kline



# Clinicians, Assessments, and Biospecimens

Dr. Jennifer Bain

Dr. Meena Balasubramanian

Dr. Billie Au

Dr. Antonie Kline

Dr. Kara Murias

Dr. Nicolo Pini

Dr. Christopher Ricupero

Dr. Barbara Corneo



Bain Brain Lab



**Volunteers!**

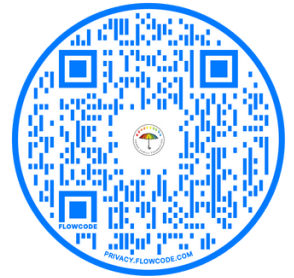
**All of the amazing families participating in person and virtually!**

# Connect With Us!

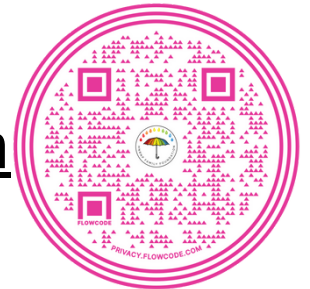
Share with #HNRNP2023



HNRNP-Related Caregiver Connect



The HNRNP Family Foundation



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[www.hnrnp.org](http://www.hnrnp.org)



[hnrnp\\_connect](https://www.instagram.com/hnrnp_connect)



# Important Contact Information

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