2023 ASXL Research Symposium
Friday, July 21 and Saturday, July 22, 2023
Simmons University
School of Business and Academic Building – 5th Floor
300 The Fenway, Boston, MA

Friday, July 21
8:30am-6:30pm

Welcome and opening
8:30-9:00am

Laura Badmaev
ARRE Foundation
Welcome and opening remarks

Stephanie Bielas, PhD
University of Michigan
Overview of the ASXL genes and their related disorders

Basic science mechanisms
9:00-10:30am

El Bachir Affar, PhD
Université de Montréal
Biochemical and functional characterization of ASXLs-BAP1 complexes

Isabella Lin, MD-PhD Candidate
UCLA-Caltech Medical Scientist Training Program
Multi-omics of Bohring-Opitz syndrome truncating ASXL1 mutations identify canonical and non-canonical Wnt signaling dysregulation

Hui Si Kwok, PhD
Harvard University
Revealing a histone methylation ceiling in EZH2-mutant lymphoma with CRISPR scanning

Animal models
11:00am-12:30pm

Svetlana Maltseva, PhD Candidate
Max Planck Institute of Biochemistry
Functional dissection of Drosophila Asx C-terminal region in vivo
Lucy Doyle, MSc, BSc
University of Edinburgh
*Understanding the function of H2A monoubiquitination in brain development*

Arneet Saltzman, PhD
University of Toronto
*Polycomb Repressive Complex 1 and histone H2A monoubiquitylation in C. elegans neuronal development*

**Therapeutic development**  
1:30-3:00pm

Yael Weiss, MD, PhD
Mahzi Therapeutics
*Drug development for rare neurodevelopmental disorders: Choices and challenges*

Ed Kaye, MD
Stoke Therapeutics
*A novel RNA-based approach to treat genetic diseases via protein upregulation*

Lavi Erisson, MD
Gensaic
*Machine guided delivery of genomic medicines*

**Natural history studies and longitudinal data**  
3:30-5:00pm

Wen-Hann Tan, BMBS
Boston Children’s Hospital
*Natural history studies: Why, how, and how not to*

Anjali Sadhwani, PhD
Boston Children’s Hospital
*Improving neurodevelopmental assessment of individuals with rare genetic conditions*

Natasha Ludwig, PhD
Kennedy Krieger Institute
*Applying the FDA Patient-Focused Drug Development Guidances to the ASXL research landscape*

**Lightning talks**  
5:00-5:30pm

Bianca Russell, MD
University of California Los Angeles
*Bohring-Opitz Syndrome EEG biomarker study*

Maya Chopra, MD
Boston Children’s Hospital
*Brain Gene Registry*
Joey Zuijdervelt, MSc
Leiden Academic Centre for Drug Research
_Early achievements in building a translational zebrafish model for Bohring-Opitz Syndrome_

Amy Brady, CNMT
University of Alabama at Birmingham
_The Noah Project_

Dale Frank, PhD
Technion
_Xenopus Laevis as a model for early neural crest development in Bainbridge-Ropers Syndrome_

**Poster session and reception**
5:30-6:30pm

**Trainee posters:**

_Zain Awamleh, PhD*
The Hospital for Sick Children
_Epigenetic dysregulation associated with pathogenic sequence variants in ASXL3_

_Isabella Lin, MD-PhD Candidate*
University of California, Los Angeles
_Multi-omics of Bohring-Opitz syndrome truncating ASXL1 mutations identify canonical and non-canonical Wnt signaling dysregulation_

_Eleanor Mills_
University of Michigan
_Understanding genetic contributors of intellectual disability: a polycomb perspective_

_Emily Peirent*_
University of Michigan
_ASXL3 links chromatin biology to neurodevelopment disorders_

_Amanda Pirin*_
University of California, Los Angeles
_Assessing pubertal timing, development, and associated factors in ASXL syndromes_

_Samantha Regan*_
University of Michigan
_Allelic and phenotypic heterogeneity in five mouse models with ASXL3 variants_

_Charles Ryan*_
University of Michigan
_Exploring the effect of ASXL3 loss of function on the H2A monoubiquitination landscape, DNA damage repair, and the cell cycle_
Additional posters:

Amy Brady, CNMT
University of Alabama at Birmingham
*Exploring PTC Read-Through Drugs for BRPS Treatment: Study on ASXL-3 Nonsense Mutations*

Amanda Johnson
ARRE Foundation
*Expanding the ASXL Research Network*

Keith McGregor, PhD
University of Alabama at Birmingham
*Advancing Therapeutics through Biotechnology Education and Innovation for Bainbridge-Ropers Syndrome*

*denotes travel grant recipient

Saturday, July 22
8:30am-12:30pm

Where we're headed: ARRE Foundation plans for the year ahead
8:30am-9:00am

Amanda Johnson
ARRE Foundation

ASXL research landscape assessment & research question ranking
9:00am-10:00am

Rachel Heilmann, PharmD
COMBINEDBrain

Collaborating for a common purpose
10:00am-10:30am

Vicky Whittemore, PhD
NIH National Institute for Neurological Disorders and Stroke
*NIH funding for rare disorders*

Workshop session: Prioritizing research needs towards translation to therapies
11:00am-12:30pm
Facilitators: Rachel Heilmann (COMBINEDBrain) and Yael Weiss (Mazhi Therapeutics)

Discussion of top-ranked research goals:

- Who else needs to be invited into the ASXL research community to meet these research goals?
- What experiments/studies, research resources, and assets are needed to meet these research goals?