

Down Syndrome Research:
The Intersection of Basic Science and Clinical Cohort Development
November 9–10, 2020
NIH-Sponsored Virtual Meeting
Times listed are in Eastern Time.

DAY 1: Monday, November 9, 2020

Session 1: Welcome and Introductions - <https://videocast.nih.gov/watch=40033>

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| 10:00–10:15 a.m. | Welcome remarks from leadership of INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome)
<i>Tara Schwetz, Associate Deputy Director, National Institutes of Health (NIH)</i>
<i>Diana Bianchi, Director, Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), NIH</i> |
| 10:15–10:45 a.m. | Summary of INCLUDE Activities and Funding
<i>Charlene Schramm, National Heart, Lung, and Blood Institute (NHLBI), NIH</i>
<i>Melissa Parisi, NICHD, NIH</i> |
| 10:45–11:15 a.m. | Keynote: perspectives from research study participants
<i>Emily Chesnut, Parent</i>
<i>Megan Bomgaars, Self-Advocate</i>
<i>Kris Bomgaars, Parent</i> |
| 11:15–11:30 a.m. | Q&A |
| 11:30–11:35 a.m. | Overview of pre-meeting working groups |

Session 2: Current State of the Science and Gaps with Regard to Basic Science and Cohort Development - <https://videocast.nih.gov/watch=40033>

Each working group will address the following questions:

1. What is the current state of the science and research gaps with regard to basic science in this domain of Down syndrome?
2. What is the current state of the science and research gaps with regard to cohort development in this domain of Down syndrome?

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| 11:35–11:50 a.m. | Neurodevelopment—structure, cognition, and language
<i>Nancy Raitano Lee, Drexel University</i>
<i>Anita Bhattacharyya, University of Wisconsin–Madison</i> |
| 11:50 a.m.–12:05 p.m. | Behavior—autism, ADHD, and regression
<i>Anna Esbensen, Cincinnati Children’s Hospital</i>
<i>Tarik Haydar, Children’s National Hospital</i> |
| 12:05–12:20 p.m. | Cardiovascular disease and pulmonary hypertension
<i>Christine Seidman, Harvard University</i>
<i>Steven Abman, Children’s Hospital Colorado</i> |
| 12:20–12:35 p.m. | Respiratory and airway conditions (including obstructive sleep apnea)
<i>Ignacio Tapia, Children’s Hospital of Philadelphia</i>
<i>Emily DeBoer, Children’s Hospital Colorado</i> |
| 12:35–12:45 p.m. | Q&A |
| 12:45–1:15 p.m. | BREAK |

- 1:15–1:30 p.m.** **Cancer—risks for leukemia and resilience to solid tumors**
Soheil Meshinchi, University of Washington
Philip Lupo, Baylor College
- 1:30–1:45 p.m.** **Autoimmunity and infections**
Bernard Khor, Benaroya Research Institute
Joaquin Espinosa, University of Colorado
- 1:45–2:00 p.m.** **Endocrine, metabolic, and skeletal conditions**
Randall Roper, Indiana University–Purdue University Indianapolis
Andrea Kelly, Children’s Hospital of Philadelphia
- 2:00–2:15 p.m.** **Aging and Alzheimer’s disease**
Benjamin Handen, University of Pittsburgh
Mark Mapstone, University of California Irvine
- 2:15–2:30 p.m.** **Q&A; charge for the breakout groups**
- 2:30–3:00 p.m.** **BREAK**

Session 3: Concurrent Breakout Sessions

3:00–4:00 p.m.

<p>Breakout Group 1: Development and Behavior https://nih.zoomgov.com/j/1613330883 Meeting ID: 161 333 0883 Passcode: INCLUDE (case sensitive) <i>Lina Patel, Children’s Hospital Colorado</i> <i>Kelly Botteron, Washington University in St. Louis</i></p>	<p>Each of these groups will address the following questions:</p> <ol style="list-style-type: none"> 1. Are there additional gaps and barriers that have not yet been raised? 2. How can these gaps and barriers be overcome?
<p>Breakout Group 2: Heart and Lung https://nih.zoomgov.com/j/1604691842 Meeting ID: 160 469 1842 Passcode: INCLUDE (case sensitive) <i>Michael Yeager, University of Colorado</i> <i>Beverly Rothermel, University of Texas Southwestern Medical Center</i></p>	
<p>Breakout Group 3: Cancer and Immunity https://nih.zoomgov.com/j/1611147879 Meeting ID: 161 114 7879 Passcode: INCLUDE (case sensitive) <i>Dusan Bogunovic, Icahn School of Medicine</i> <i>Kelly Sullivan, University of Colorado</i></p>	
<p>Breakout Group 4: Aging and Metabolic Conditions https://nih.zoomgov.com/j/1609379034 Meeting ID: 160 937 9034 Passcode: INCLUDE (case sensitive) <i>Ann Cohen, University of Pittsburgh</i> <i>Sheela Magge, Johns Hopkins University</i></p>	

4:00–4:15 p.m. **BREAK**

Session 4: Breakout Reports and Joint Basic and Cohort Development Session -

<https://videocast.nih.gov/watch=40033>

- 4:15–5:15 p.m.** **Breakout group report-backs**
- 5:15–5:30 p.m.** **Discussion**
- 5:30 p.m.** **Closing comments**
Gary Gibbons, Director, NHLBI

DAY 2: Tuesday, November 10, 2020

Session 5: Joint Basic and Cohort Development Session - <https://videocast.nih.gov/watch=40034>

10:00–10:10 a.m. Welcome remarks from INCLUDE leadership
10:10–10:20 a.m. Charge for the day

Session 6: Concurrent Sessions

10:20–11:50 a.m. State of the science: basic science and cohort development

<u>Basic Science</u>	<u>Cohort Development</u>
Model systems and tools to advance Down syndrome basic and preclinical science https://nih.zoomgov.com/j/1613762076 Meeting ID: 161 376 2076 Passcode: INCLUDE (case sensitive)	INCLUDE Data Coordinating Center and existing and future cohorts https://nih.zoomgov.com/j/1606299989 Meeting ID: 160 629 9989 Passcode: INCLUDE (case sensitive)
Introduction <i>Roger Reeves, Johns Hopkins University</i>	Introduction to the INCLUDE Data Coordinating Center <i>Joaquin Espinosa, University of Colorado</i>
Current state of mouse models and phenotypic drift <i>Randall Roper, Indiana University–Purdue University Indianapolis</i>	Overview of existing cohorts <i>Amy Brower, American College of Medical Genetics and Genomics</i>
TcMAC21 and upcoming rat models <i>Roger Reeves, Johns Hopkins University</i>	Considerations for GUIDs and linkages <i>Russ Waitman, University of Missouri–Kansas City</i>
Current state of human stem cell research for DS <i>Anita Bhattacharyya, University of Wisconsin–Madison</i>	Biospecimen collection and associated datasets <i>Elizabeth Head, University of California Irvine</i>
Extracellular vesicles and deposition of amyloid in organoids derived from iPSCs <i>Vasiliki Machairaki, Johns Hopkins University</i>	

11:50 a.m.–12:15 p.m. BREAK

Session 7: Concurrent Breakout Sessions

12:15–1:45 p.m.

Breakout Group 5: Ensuring Robust iPSC and Organoid Systems as Preclinical Models https://nih.zoomgov.com/j/1608849481 Meeting ID: 160 884 9481 Passcode: INCLUDE (case sensitive) <i>Peng Jiang, Rutgers University</i> <i>Jeanne Lawrence, University of Massachusetts Medical School</i>	Each of these groups will address the following questions: <ol style="list-style-type: none">1. How does one choose the best model system?2. How does one ensure rigor and reproducibility?3. What clinical and epidemiological data would be useful to guide basic science studies?4. What biospecimens would basic scientists like to have access to and for what uses?
Breakout Group 6: Ensuring Robust Animal Model Systems as Preclinical Models https://nih.zoomgov.com/j/1616635653 Meeting ID: 161 663 5653 Passcode: INCLUDE (case sensitive) <i>Benjamin Tycko, Hackensack University</i> <i>Cathleen Lutz, The Jackson Laboratory</i>	
Breakout Group 7: Clinical Phenotyping and Minimal Common Data Elements https://nih.zoomgov.com/j/1616222412 Meeting ID: 161 622 2412 Passcode: INCLUDE (case sensitive) <i>Maria Stanley, University of Wisconsin Madison</i> <i>Nicole Vasilevsky, Oregon Health & Science University</i>	Each of these groups will address the following questions: <ol style="list-style-type: none">1. What clinical and phenotyping data should be collected?2. What biospecimens should be collected and for what purposes?3. What fundamental scientific questions are addressable by large cohort studies?4. What clinical scenarios require a deeper understanding of the underlying science?
Breakout Group 8: Biospecimens and Related Omics Datasets https://nih.zoomgov.com/j/1609959633 Meeting ID: 160 995 9633 Passcode: INCLUDE (case sensitive) <i>Adam Resnick, Children's Hospital of Philadelphia</i> <i>Qiang Chang, University of Wisconsin-Madison</i>	

1:45–2:00 p.m.

BREAK

Session 8: Breakout Reports and Joint Basic and Cohort Development Session -

<https://videocast.nih.gov/watch=40034>

2:00–3:00 p.m.

Breakout group report-backs

3:00–3:40 p.m.

Panel discussion:

What do clinical investigators want basic scientists to know?

What do basic scientists want clinical investigators to know?

Nicole Baumer, Boston Children's Hospital

George Capone, Kennedy Krieger Institute

Roger Reeves, Johns Hopkins University

Christine Seidman, Harvard University

Stephanie Sherman, Emory University

Maria Stanley, University of Wisconsin–Madison

3:40–4:00 p.m.

Wrap-up and next steps

4:00 p.m.

Close of Day 2