2024 Rare Disease Conference
February 29 - March 1, 2024
Schedule of Events

Thursday, February 29 — **University of Notre Dame**

Thursday, 29 February

4:00 - 5:00 PM Jordan Hall of Science, Room 105	**Plenary Talk: Accelerating new therapies for rare diseases: The Oxford Harrington Rare Disease Centre** Matthew Wood, M.D., Ph.D., University of Oxford
5:00 - 6:15 PM Jordan Hall Galleria	**Welcome Reception & Rare Disease Day Dinner; Presentation of Student Awards**
6:30 - 6:45 PM Hesburgh Library	**Light Up For Rare Ceremony** Prayer led by Fr. Pete McCormick, C.S.C.
7:00 PM Purcell Pavilion	**Women's Basketball Game vs. Virginia Tech**

Friday, March 1 — **Embassy Suites at Notre Dame, 1140 E. Angela Blvd.**

Friday, March 1

8:00 - 9:00 AM	**Registration and continental breakfast**
9:00 - 9:30 AM	**The role of cardiovascular genetics in the diagnosis and management of rare disease** Stephanie Ware, M.D., Ph.D., Indiana University School of Medicine
9:30 - 9:50 AM	**Tissue engineered disease models for rare disease research** Pinar Zorlutuna, Ph.D., University of Notre Dame
9:50 - 10:20 AM	**Unmasking the selective functions of extracellular signal-regulated kinase-1/2 activation in the developing nervous system** Jason Newbern, Ph.D., Arizona State University
10:20 - 10:40 AM	**Human neurons: A tool for understanding disease mechanisms of intellectual disability** Chris Patzke, Ph.D., University of Notre Dame

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BOLER-PARSEGHIAN
Center for Rare & Neglected Diseases

10:40 - 11:10 AM **Panel: Rare neurological diseases**
Jason Newbern, Ph.D., Arizona State University;
Matthew Wood, M.D., Ph.D., University of Oxford;
Tom Hamilton, Board Member, Friedreich's Ataxia Research Alliance
Moderated by Sean Kassen, Ph.D, University of Notre Dame

11:10 - 11:15 AM **Testimonial: Brandi Wampler**
Parent of a child with a rare disorder (ARHGEF9 gene mutation)

11:15 - 12:10 PM **Lunch**

12:10 - 12:30 PM **Finding new treatments for rare cancers with genetic modeling and mechanism studies**
Xin Lu, Ph.D., University of Notre Dame

12:30 - 12:50 PM **Immunomechanics in rare cancers**
Meenal Datta, Ph.D., University of Notre Dame

12:50 - 1:20 PM **Panel: The genetics of rare cancers: Redefining what is rare**
Xin Lu, Ph.D., University of Notre Dame;
Meenal Datta, Ph.D., University of Notre Dame;
Zach Schafer, Ph.D., University of Notre Dame
Moderated by Sharon Stack, Ph.D., University of Notre Dame

1:20 - 1:25 PM **Testimonial: Hannah Gunn**
Current student (ND '27) with family member with TNF receptor-associated periodic fever syndrome

1:25 - 1:35 PM **Break**

1:35 - 2:05 PM **Defining biomarkers in Niemann-Pick Type C Disease**
Stephanie Cologna, Ph.D., University of Illinois Chicago

2:05 - 2:35 PM **Panel: Using biomarkers to predict NPC disease progression**
Stephanie Cologna, Ph.D., University of Illinois Chicago;
Elizabeth Berry-Kravis, M.D., Ph.D., Rush University;
DeAnna Odeh , Parent to a child with Niemann-Pick Type C;
Kevin Vaughan, Ph.D., University of Notre Dame
Moderated by Sean Kassen, Ph.D., University of Notre Dame

2:35 - 2:40 PM **Testimonial: Cathleen Reisenauer**
Parent to two children with Glycogen Storage Disease Type III

2:40 - 3:10 PM

A decade of rare disease program experience at Children's Wisconsin/Medical College of Wisconsin
Donald Basel, M.D., Medical College of Wisconsin

3:10 - 3:50 PM

Panel: Redefining rare disease
Donald Basel, M.D., Medical College of Wisconsin;
Stephanie Ware, M.D., Ph.D., Indiana University Medical School;
Santiago Schnell, D.Phil., University of Notre Dame
Moderated by Sean Kassen, Ph.D., University of Notre Dame

4:00 - 6:00 PM

Reception