

Friday, February 9, 2018

Jordan Hall Reading Room 10am-5pm



Rare Disease Day®

Rare Disease and Brain

10:30-10:40am

Introduction Mr. Rob Long (brain cancer survivor)

10:40-11:00am

Siyuan Zhang, M.D., Ph. D., Dee Assistant Professor, Dept. of Biological Sciences, ND

Rare Breast Cancer to Brain Metastasis

11:00-11:20am

Andrew Lieberman, M.D., Ph. D. Gerald D. Abrams Collegiate Professor, University of Michigan

Identification of targets and therapeutic strategies for Niemann-Pick disease

11:20-11:40am

Johan VanHove, M.D., Ph. D., University of Colorado
Clinical NKH

11:40-11:50am

NKH Patient discussion panel

Hematological and Respiratory Disease

11:50-12:00noon

Introduction Elena Kolarevic (Living with anemia)

12:00- 12:20pm

Mohandas Narla, DSc. Member New York Blood Center, NY
Inherited Anemias

12:20-12:40pm

Jeff Schorey Ph. D., George B. Craig Professor, Dept. of Biological Sciences, ND
Cystic Fibrosis and non-tuberculosis mycobacteria

12:45-1:30pm

LUNCH

Models in Rare Diseases

1:30-1:40pm

Introduction Karen Quandt (NPC Patient family)

1:14-2:00pm

Suhail Alam Ph. D., Research Assistant Professor, Dept. of Biological Sciences, ND
Chronic HDACi therapy to treat lung disease in NPC

2:00-2:20 pm

Crislyn D'Souza-Schorey Ph. D., Pollard Professor and Dept. Chair
Biological Sciences, ND
Molecular Properties of Rare Melanomas

2:20-2:40pm

Shaun Lee, Ph. D., Monahan Family Associate Professor, Dept. Biological Sciences, ND
Protein folding and Predicting Disease Severity

2:40- 3:00pm **Kevin Vaughn**, Ph. D., Professor, Dept. Biological Sciences, ND
StarD9 and neurodegeneration in a mouse model

3:00-3:20pm **Xin Lu**, Ph. D., John M. and Mary Jo Boler Assistant Professor, Dept. of
Biological Sciences, ND
Targeting tumor microenvironment to treat penile cancer

3:20 pm-3:50 pm **Teaser Talks 1-3 min: Posters/Booths Connecting Research
and Patients**

- ND Student Clubs: Make-A-Wish/RareND/Invisibilities/UpliftingAthletes/Comp. Care.
- HematologicalandRespiratoryDiseases
- Rare Tumor Biology
- IntellectualDisabilities
- Neurological Diseases
- Musculoskeletal and other Rare Disorders
- Global Genes

3:50-5:00 pm **POSTER/BOOTHS SESSION (Also Continued on Saturday)**

5:00-7:00pm **Dinner**
Welcome (2-5 min)
Nicole Boice, CEO and Founder, Global Genes
*RARE Patient Advocates are Rock stars - The Increasing Role of Patients
As Health Care Architects, Investors and Entrepreneurs (20 min)*

Nicole is CEO/Founder of Global Genes. Since 2008 Nicole has been helping families affected by rare diseases connect with tools, resources, and support to help eliminate challenges in rare diseases. Global Genes is a multimillion dollar nonprofit organization that has positively impacted thousands of patients worldwide through development of educational tools and resources and hosting world class patient focuses events. Nicole has an extensive business, sales, marketing and consulting background and has worked in media, pharmaceutical, venture and high-tech sectors. Nicole will speak about the roles that patients play to positively affect disease related outcomes.

Rob Long, Director of Strategic Development, Uplifting Athletes
Living the Rare Life Full Circle (20 min)

Rob is the Director of Strategic Development for Uplifting Athletes. Rob is a survivor of anaplastic astrocytoma, a rare brain cancer. He graduated from Syracuse University, was a 4 year starter/punter for the Orangemen Football Team and was voted by his teammates as Captain for two of those years. Rob was on his way to becoming a punter for the NFL when he was given his rare diagnosis. He went on to receive his Bachelor of Science from the Martin J Whitman School of Mgmt and a masters from SI Newhouse School of Public Communications. Currently, Rob oversees 25 chapters of Uplifting Athlete networks in the US consisting of college/athletes. Rob is here to share his inspiring story of survival and his work with Uplifting Athletes.

7:30pm **Hockey Game: Notre Dame vs. Ohio State**

Rare and Neglected Disease Day Conference

Saturday, February 10, 2018

Jordan Hall 105 8:30 am-5 pm



Rare Disease Patient Research and Advocacy

- 8:30-8:45am** Welcome, University of Notre Dame Glee Club
- 8:45-9:05am** Elizabeth Berry-Kravis, M.D., Ph. D., Rush Medical Center
Update on Niemann Pick Type C (NPC) Clinical Trial
- 9:05-9:15am** NPC patient panel discussion led by **Karen Quandt**
- 9:15-9:35am** Russel Ware, M.D., Ph. D., Cincinnati Children's Hospital
Sickle Cell Therapies
- 9:35-9:45am** Sickle Cell patient panel discussion led by **Gary Gibson**
- 9:45-10:00am** Short Coffee Break
- 10:00-10:20am** Kasturi Haldar, Ph. D., Director, Boler-Parseghian CRND
Genetic and Epigenetic Treatments for Rare Diseases (NPC, NKH and others)
- 10:20-10:30am** Patient Panel
- 10:30-10:50 am** John Crowley, Chairman and CEO, Amicus Therapeutics
New Technologies and Advances in Rare Diseases across Industry
- 10:50-11:00 am** Megan Crowley, ND '2019.
Growing up knowing your father is fighting to save your life against a rare disease
- 11:00am-11:30pm** Teaser Talks 3 min: Posters/Booths Connecting Research and Patients
- ND Student Clubs: Make-A-Wish/RareND/Invisibilities/UpliftingAthletes/Comp. Care
 - Hematological and Respiratory Diseases
 - Rare Tumor Biology
 - Intellectual Disabilities
 - Lysosomal Storage Disorders
 - Neurological Diseases
 - Musculoskeletal and Other Rare Disorders
 - Global Genes
- 11:30-3:00pm** Lunch/Posters/Booths Connecting Research and Patients.
- 3:00-4:30pm** Patient/students partners present 9 rare diseases
- 3:00-3:10pm** Neurofibromatosis Type 1 (Brian Crowley and Joseph Stevens)
Nicole Sevison
- 3:10-3:20pm** Rett Syndrome (Daniel Hogan and Kayla Perez)
David Faverty

3:20-3:30pm	NKH/ Liver Cancer (Emily Casaletto and Meghan Cohoon) Amanda Almany
3:30-3:40pm	SGS (Gabriela Moro) Melissa and Tim Kern
3:40-3:50pm	Fragile X (Monica Bell and Jori Waner) Sarah Snyder
3:50-4:00pm	Angelman (Lucie Ly and Nicole Zolman) Deanna Jeffrey
4:00-4:10pm	Turner Syndrome (Cecilia Barry and Meredith McClain) Madeline Moeller
4:10-4:20pm	Diamond Blackfan Anemia (Simon Padanilam) Elena Kolarevic
4:20-4:30pm	Sickle Cell- Anemia (Courtney Koch) Antoinetta Freeman
4:30-4:40pm	Desmoplastic Medulloblastoma (Frances Neunuebel)
4:45pm	Closing Remarks



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