Rare and Neglected Disease Day Conference  
Friday, February 9, 2018  
Jordan Hall Reading Room 10am-5pm

**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*

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

- ND Student Clubs: Make-A-Wish/RareND/Invisililies/UpliftingAthletes/Comp. Care.
- Hematological and Respiratory Diseases
- Rare Tumor Biology
- Intellectual Disabilities
- Neurological Diseases
- Musculoskeletal and other Rare Disorders
- Global Genes

3:50-5:00pm  **POSTER/BOOTHES 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
Welcome, University of Notre Dame Glee Club
8:30-8:45am

Elizabeth Berry-Kravis, M.D., Ph. D., Rush Medical Center
Update on Niemann Pick Type C (NPC) Clinical Trial
8:45-9:05am

NPC patient panel discussion led by Karen Quandt
9:05-9:15am

Russel Ware, M.D., Ph. D., Cincinnati Children’s Hospital
Sickle Cell Therapies
9:15-9:35am

Sickle Cell patient panel discussion led by Gary Gibson
9:35-9:45am

Short Coffee Break
9:45-10:00am

Kasturi Haldar, Ph. D., Director, Boler-Parseghian CRND
Genetic and Epigenetic Treatments for Rare Diseases (NPC, NKH and others)
10:00-10:20am

Patient Panel
10:20-10:30am

John Crowley, Chairman and CEO, Amicus Therapeutics
New Technologies and Advances in Rare Diseases across Industry
10:30-10:50 am

Megan Crowley, ND ‘2019.
Growing up knowing your father is fighting to save your life against a rare disease
10:50-11:00 am

Teaser Talks 3 min: Posters/Booths Connecting Research and Patients
- ND Student Clubs: Make-A-Wish/RareND/Invisililies/UpliftingAthletes/Comp. Care
- Hematological and Respiratory Diseases
- Rare Tumor Biology
- Intellectual Disabilities
- Lysosomal Storage Disorders
- Neurological Diseases
- Musculoskeletal and Other Rare Disorders
- Global Genes
11:00am-11:30pm

Lunch/Posters/Booths Connecting Research and Patients.
11:30-3:00pm

Patient/students partners present 9 rare diseases
3:00-4:30pm

Neurofibromatosis Type 1 (Brian Crowley and Joseph Stevens)
Nicole Sevison
3:00-3:10pm

Rett Syndrome (Daniel Hogan and Kayla Perez)
David Faverty
3:10-3:20pm
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