Rare 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)

1:40-2:00pm  
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/Invisililies/UpliftingAthletes/Comp. Care.
- Hematologic Disorders
- Rare Tumor Biology
- Intellectual Disabilities
- Lysosomal Storage Disorders
- Neurological Diseases
- Fibromas and other disorders
- Global Genes

3:50-5:00 pm  **POSTER/BOOTH 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 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/Invisililies/UpliftingAthletes/Comp. Care  
- Hematologic Disorders  
- Rare Tumor Biology  
- Intellectual Disabilities  
- Lysosomal Storage Disorders  
- Neurological Diseases  
- Fibromas and other 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

3:10-3:20pm  
NKH/ Liver Cancer

3:20-3:30pm  
SGS
3:30-3:40pm  Fragile X
3:40-3:50pm  Angelman
3:50-4:00pm  Turner Syndrome
4:00-4:10pm  DiGeorge Syndrome
4:10-4:20pm  Diamond Blackfan Anemia
4:20-4:30pm  Sickle Cell- Anemia
4:45pm  Closing Remarks