The Boler-Parseghian Center for Rare and Neglected Diseases is deeply grateful for an extraordinary weekend celebrating our 10th Annual Conference Advancing Rare Disease Research, Therapy and Patient Advocacy on March 1-2 2019!

It was our largest meeting with over 200 in attendance. Thanks to our illustrious alumni, outstanding faculty, students, staff, patient families, clinical partners and other community partners for making it a great success!

The Friday March 1 dinner, was introduced by Dr. Kasturi Haldar (Director, CRND) and Mrs Barbara Calhoun (Outreach Coordinator CRND). Dean Mary Galvin (Dean, College of Science, COS) spoke to the commitment of the COS and the University to Rare and Neglected Diseases and led with the invocation. The keynote speaker Mr. Matt Boler, Chairman of the Board, President and CEO Boler Company inspired us with sharing his family’s pledge to this area of high unmet need and potential for research discoveries.
We were also excited to establish two new awards for our exceptional undergraduates. The first was the **Megan K. Crowley Award for Patient Advocacy**, whose recipient Megan Crowley, ND ’19 is a tireless patient advocate and President of the Notre Dame Chapter of Make a Wish Foundation that aims to grant a wish of every child diagnosed with a life threatening condition. CRND is honored Megan came to study at Notre Dame.

The second award was the **John M. and Mary Jo Boler Rare Disease Research Award** in honor of the parents of Matt Boler and Jill McCormack and the resonance of their generous philanthropy with rare diseases.

The recipient Gabrielle O’Dougherty ND ’19 in partnership with a local patient family and their neurosurgeon developed a unique case report on Shprintzen Goldberg Syndrome (SGS) that is currently under revision for publication. O’Dougherty et al, *Case Report: Complications of Insufficient Dura and Blood Loss during Surgical Intervention in Shprintzen-Goldberg Syndrome.*

Jill McCormack, Trustee Boler Family Foundation, presented both awards.

We completed Friday evening with a rousing hockey game honoring patients and families. Notre Dame came from behind to beat Penn State 5-4!
Saturday March 2 morning began with a dazzling Glee Club performance of the Rare Disease Fight Song and gracious welcome Deputy Chief of Staff Suzanna Fitzberg, South Bend Mayor’s Office.

Dr. Xin Lu, John M and Mary Jo Boler Assistant Professor, Boler-Parseghian Center delivered the keynote lecture on preclinical models that help move research findings from the laboratory to treatments in the clinic for rare tumors and cancers.

This was followed by three panel discussions with active audience participation! In the first panel on ‘Rare Disease Research and Therapy’, John Crowley (CEO Amicus), Steve Aselage (Board Member Retrophin), Dr. Diana Wetmore (V.P. and Director Harrington Institute), Dr. Elizabeth Berry-Kravis (Rush Medical Center) joined Drs. Brian Blagg (Director Warren Center, Notre Dame) and Kasturi Haldar (Director CRND) to discuss the promises of partnerships between pharmaceutical industry and academic researchers and the importance of patient advocates to accelerate new treatments for rare diseases. They also reviewed challenges and how to bridge them with new models of research and development in the 21st century.

The second panel on ‘Resources and Empowerment’ comprised of ND faculty Drs. Suhail Alam, Rebecca Wingert, John Koren and Siyuan Zhang, as well as Kunigunda Szentes from the IDEA Center at ND. The panel also included patient advocates Gillian Shaw (Enfocus) and Nicole Boyce (Founder Global Genes). Drs. Alam and Wingert presented on strengths of murine neurological disorders and zebrafish genetic models available at the Freimann Life Sciences Center.
Dr. Koren has newly established a Biological Screening Core at the Warren Center for Drug Discovery. Dr. Zhang emphasized the importance of computational training to understand a complex target or drugs directed to it, to fully understand the properties of new medicines, while Ms. Szentes outlined funding mechanisms made available to move them to market. Gillian Shaw and Nicole Boyce are key community stakeholders who broadly empower patients in activities that range from home-based technology-assisted disease management to building patient services organizations (PSO’s) that effectively fund-raise for research and treatment in rare diseases.

The third panel on Saturday morning was on ‘Patient Health & Advocacy’. Dr. Mary Alice Reid, (Beacon Pediatrics), Karen Quandt, R.N., (NPC Patient Family & Advocate), Tarah O’Sullivan & Mary Fitzgerald (NKH Patient Advocates), Dr. Daniel Fulkerson, (Beacon Health), Francis Fitzgerald (ND’18) and Rob Long, (Uplifting Athletes), spoke to how they as clinicians and patients came to be advocates and address daunting tasks of satisfactory and timely diagnosis and care for rare disease patients. The panel concluded there may be need for post baccalaureate training in patient advocacy to support families to access technology-driven treatments and services as these become increasingly available in many rare diseases.
The Poster session overlapped with lunch from 12.30-2.00 pm. To draw attention and audience students utilized one-minute teaser talks to advertise 26 posters assembled into nine subject areas of Neurological disorders, Hematological and Respiratory Diseases Rare Tumor Biology, Intellectual Disabilities, Lysosomal Storage Disorders, Musculoskeletal and other rare disorders, Rare ND & Student Clubs, Global Genes and Uplifting Athletes.

Last but most importantly were presentations by patient families on their personal natural histories of Spinal Muscular Atrophy with Respiratory Distress, Angelman Syndrome, Von Hippel Lindau Disease, Leber's Hereditary Optic Neuropathy, Coats Disease, Rett Syndrome, Neurofibromatosis Type 1, Mast Cell Activation Syndrome/ Eosinophilic Esophagitis, Sickle Cell Disease, Shprintzen Goldberg Syndrome and Non-ketotic Hyperglycinemia. We were transfixed by their courage, grace and tenacity to fight and raise awareness that rare diseases are not strange afflictions but rather part of our common genetic heritage and at an overall prevalence of 1 in 10, a forefront of human wellness.

Once again, we thank all the participants for their invaluable contributions. We thank Corianne Kellems for organizational support and Matt Frazier for photography. Finally, we give a shout out to Haldar lab members, Niraja Suresh, Arpitha MysoreRajashekar, Innocent Safeukui, Suhail Alam, Joe Farris, Katharyn Lee, Morgan Hiller, Abhishek Trivedi, Maisha Khair Nima and Fang Liu for their many hands and team spirit to bind the weekend.

Kasturi Haldar and Barbara Calhoun, CRND 2019.
Patient Presenters

Frances Fitzgerald

Chris Kiergan

Melissa, Tim Kern

Sarajane Roenke

David Faverty

Elena Kolarevic

Tim Ponisiak

Nicole and Skylar Sevison

Megan and Ben Webber ‘know the Glow’

Trevor and Antoinetta Freeman

Greg and Amanda Almany

ND-NKH patient team

Haldar Lab