



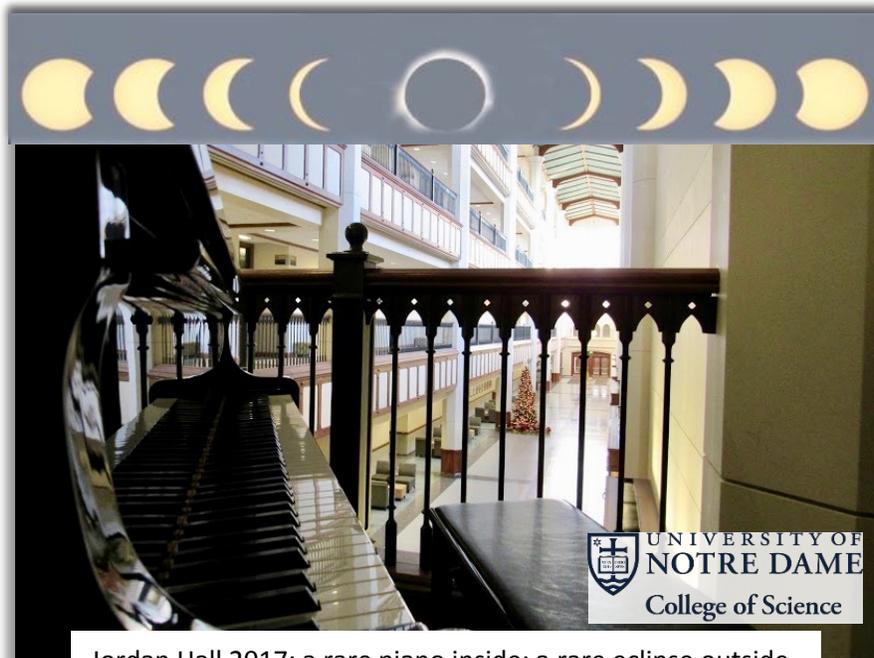
BOLER-PARSEGHIAN

Center for Rare & Neglected Diseases

Signal CRND Newsletter Vol. 6, 2016-2017

Mission Statement. The Boler-Parseghian Center for Rare and Neglected Diseases (CRND) at the University of Notre Dame is charged with the mission of supporting research of Notre Dame faculty in developing drug and additional therapies for rare and neglected diseases.

2016-2017 saw CRND's web site transition from a 'live blog' to a structured portal shepherded by Ms. Corianne Kellems, who joined us in March 2017. It reflects our programmatic growth, starting from laboratory research in one neurological disease in 2008 to more than ten different rare genetic diseases in 2017 that synergize with Notre Dame's existing strengths in neglected infectious diseases. Natural history studies have also significantly expanded to 15 diseases. The credit goes to dynamic faculty, fellows and students, patients, clinical and pharma partners and their interdisciplinary research that pushes the boundaries of genetic and precision medicine to replace devastating disease.



Jordan Hall 2017; a rare piano inside; a rare eclipse outside.

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Rare and neglected diseases 2016-2017

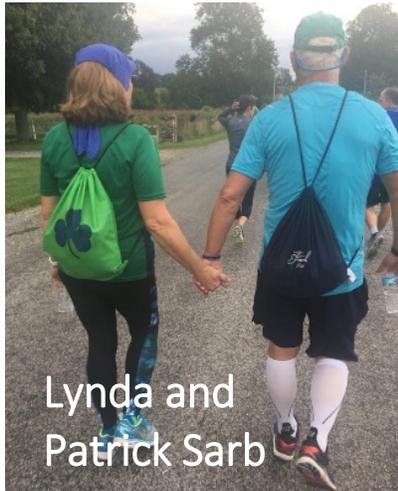
A rare disease is defined as one where the incidence is less than 200,000 in the US. Across 7,000 diseases, the chances of being affected by a rare disease is 1 in 10 worldwide. Therefore, over a lifespan, the chance of being afflicted is not that rare, albeit always unexpected.

All rare diseases impact our lives. Patients and caregivers encounter daily struggles for many years in a health system that generally poorly understands the crippling disease. The terrain is uncharted, uphill and takes herculean determination propelled by faith. Patrick Sarb ('73), Lynda Sarb ('73) and Sean Nohelty ('97) were pilgrims in the steps of Father Sorin on the ND trail in August 2017. Their mission was to raise awareness and research funds for Non Ketotic Hyperglycinemia. We are also privileged to have joined with leading rare disease advocates, additional ND families, Centers and Institutes to expand areas of rare and neglected disease research to find cures that afflict local and global communities.

We are excited to announce that the 2018 Rare and Neglected Disease Conference will kick off with keynotes from Nicole Boyce, Founder and CEO, Global Genes and Rob Long, Director of Uplifting Athletes (who beat the rare cancer that prevented his NFL career). We will celebrate our rare disease champions who fight rare and neglected diseases as patients, students, researchers and clinicians. Join us with ND hockey! Everyone is welcome but registration needed. [Click here for registration](#)

All pictures in this newsletter are shown with permission.

2. Notre Dame Trail and Non Ketotic Hyperglycinemia



Lynda and Patrick Sarb



Sean Nohelty

The ND Trail pilgrimage (Aug. 13-26, 2017) commemorated the 175th anniversary of Notre Dame's founding by Holy Cross Priest Fr. Edward Sorin. Patrick Sarb ('73) retraced steps of Fr. Sorin, from Vincennes, Indiana to Notre Dame, a distance of over 300 (see map). His wife Lynda Sarb ('73) joined him for the final three days and

40 miles of the ND Trail. Sean Nohelty ('97) walked the trail for five days and 67 miles. We honor the participation that deepened their ties to Notre Dame and raised awareness to support therapies for Non Ketotic Hyperglycinemia (NKH), a rare metabolic disorder that causes severe neurodevelopmental disease.

Pat and Lynda established the 'ND-NKH' fund in honor of their grandson Owen Sarb diagnosed with NKH within months of birth and now, against all odds is age 5.

Sean's sister and brother-in-law Mary and John Fitzpatrick sadly lost their daughter Fiona to NKH in 2009 when she was three years old. In her memory, they established 'Fighting for Fiona and Friends' (FFF).

For Sean and Pat's family histories and trail reflections see:



https://crnd.nd.edu/assets/258860/reflections_along_the_notre_dame_trail.pdf

<https://trail.nd.edu/news/day-one/>

<http://www.wndu.com/content/news/A-Pilgrims-Purpose-440975343.html>

<http://trail.nd.edu>

CRND News

CRND is also grateful for the response and support of the global NKH patient community in fundraising and participation in research. Thank you to all who have supported ND research including FFF, Sarb families, Almany family, Judy Alvarez and BEE-lievers as well as the many who generously contributed to ND Day. Together they helped raise almost \$90,000 for NKH research in 2016-2017! Support has enabled the use of a leading genetic methodology CRISPR-Cas9 to identify ways to treat NKH patient cells with new therapies developed by CRND. CRISPR-Cas9 was also used to develop a NKH mouse colony to translate therapies in mouse models. We continue to work with Dr. Van Hove and his colleagues on 'metabolomics' and have provided Dr. Van Hove support for this work.



Judy Alvarez and BEE-lievers



Amanda, Nora, and Andrew Almany



Rare Disease Patient Advocacy & Research.

Celebrating Champions of Rare, Neglected & Invisible Diseases



ND Alum John Crowley and his family's story of refusal to accept their children's diagnosis of 'Pompe', a rare fatal disorder, and developing a therapy to save them is well chronicled in the best-selling book 'The Cure' and movie 'Extraordinary Measures'. Mr. Crowley, Chairman and CEO of Amicus Therapeutics Inc. has been a tireless champion for patient communities. On Nov. 17th, he visited to share his intrepid path and advised multiple groups of ND undergraduates who are emerging advocates for vulnerable patients. RareND, Make A Wish, Invisillies, Compassionate Care and Uplifting Athletes along with BIOS 60565 students presented summaries as indicated below.

RareND educates and raises awareness with rare disease patients, their service organizations, caregivers and researchers (on-campus patient engagement is supported by Nurse Calhoun, CRND). They volunteer in local clinics with medically fragile children. Check out the full range of activities on Facebook (Rare ND)!

The Invisillies is an all women group who suffer from illnesses without external or obvious symptoms, and therefore 'invisible' illnesses, which results in public

disbelief and alienation. Invisillies strives to change this. The first Invisible Illness Awareness Day was a great educational success. Look out for a repeat in 2018!

Uplifting Athletes is a nationwide network of chapters led by college football student-athletes, who underwent expansion of activities and started ND chapter of the very first Lift for Life, the signature fundraising event for Uplifting Athletes, to raise \$17,457.65 (crushing their goal of \$10,000).

Make a Wish is a national organization with the goal of supporting the wishes of children with life-threatening medical conditions. Members of the ND Make A Wish Club shared ways in which members were successful in raising funds to grant wishes to chronically ill children.

Compassionate Care in medicine is a primary focus of the Ruth M. Hildebrand Center at ND. A new opportunity available is Nava Health, a program that checks in with low Socioeconomic Status (SES) patients to ensure proper compliance with medicines.

BIOS 60565 CRND students assist with defining natural histories of rare diseases with local and global rare disease patient and clinical partners. 200 student milestone anticipated in Spring 2018. Some are Editorial Interns for National Organization of Rare Disease's 3000+ Reports Database.

John Crowley suggested that student groups should combine efforts on campus events and activities. For example, the Make A Wish Foundation can assist with promoting events that can benefit all groups. Global Genes, a world leader in rare communication/awareness may have resources for students. Rare ND 'ZEBRA,' a phone app to assist rare disease patients with complex medication and other schedules, improved their path to a sponsor.

Another powerful message from John Crowley to the students was that they should focus on who people truly are, on the inside. This has special meaning for Rare Disease patients whose conditions and disease progression are often poorly understood. Their fighting spirits that overcome disabilities enlighten all who believe in hope, equity, action, policy, awareness and funds for research and care to advance treatments for rare disease patients.

Global Impact of Rare Diseases in Our Community: Neurofibromatosis Workshop, Jordan Hall ND - Sept 23rd, 2017

CRND's engagement with Neurofibromatosis (NF) began in 2015 through pediatrician **Dr. Mary Alice Reid** (ND '84,) and her patient family (mother Nicole Sevison with daughter Skylar) when they were invited to campus by RareND club. Skylar was diagnosed with an unusual case of NF1, which led to the development of case report (co-led by Dr. Reid and Senior Madeline Zupan ND '16, ESTEEM '17; doi:



Dr. Mary
Alice Reid
ND '84



Skylar
Nicole Sevison

<http://dx.doi.org/10.1101/066316>) suggesting that current federal guidelines for NF1 diagnosis should be expanded, exemplifying how a single local case of a rare disease may have broad impact. *Supported by Ganey from the Center for Social Concerns, UND.*

In 2017, Alec Biscopink (ND '18) has undertaken Senior Thesis work in NF1. He is utilizing the resources of the Michiana Health Information Network (MHIN) (one of the largest health information exchanges in the country with ~80,000 rare disease records) as well as the published literature of ~1400 international patients to further analyze the global significance of Skylar Sevison's unique case.



Diana
Haberkamp



Liz
Campana



Alec presented his preliminary findings at the NF1 workshop held by CRND and the Neurofibromatosis Midwest Network, whose focus was education, patient support and awareness. Diana Haberkamp and Liz Campana, from NF Midwest Network shared resources on both clinical and research findings with BIOS 60565 and local NF1 patients and Dr. Reid. Skylar attended too - improved and walking, with a brace!



All students are trained in patient privacy and are HIPAA (Health Insurance Portability and Accountability Act) compliant. Patient engagement is supported by Nurse Calhoun, CRND

Neuroscience and Behavior

BIOS 40450 Developing Health Networks in Rare and Neglected Neurological Disorders offered in Winter-Spring semester, fulfils a science requirement for Neuroscience and Behavior major. The course engages upper level undergraduate (and graduate) students in clinical research in rare, neurological diseases through direct examination of patient medical records that have been contributed to CRND by families who suffer from these diseases. The class covers human genetics and molecular mechanisms and links to clinical progression as a cumulative disease score.

BIOS 60565 offered in Fall is more exploratory to beta test research opportunities on intersection of neurobiology and epigenetics (such as Kabuki and Rett Syndromes) and serve as portals to broadly understanding intellectual disability and autism spectrum disorders.



On Dec 1st, 2017 BIOS 60565 was privileged to host a Rett Syndrome patient family. As a final take home assignment, students researched how autism spectrum disorders (ASDs) differ from Rett Syndrome and when a Rett patient may also be considered to meet criteria of ASD.

All students are trained in protecting patient privacy and HIPAA (Health Insurance Portability and Accountability Act) compliance. All patient engagement is supported by Nurse Calhoun, CRND. In 2017 Nurse Calhoun also supervised 8 juniors and seniors undertaking individual projects in natural history of rare neurological diseases that include other diseases of intellectual disability such as Kabuki syndrome.



Nurse Calhoun also provided clinical perspective for a neurodegeneration lab led by Dr. Suhail Alam (Research Assistant Professor and Director CRND External programs) for students in Dr. Nancy Michael's Neuroscience and Behavior lab 21450.

ND-NORD Editorial Internships

ND students have been partnering as Editorial Interns with the National Organization for Rare Diseases (NORD) to help them continuously update a publication data base of rare disease summaries. The summaries are used by families afflicted with a rare disease, to learn about its genetic basis and progression, physicians and centers that treat and study the disease, as well peer groups and funding opportunities that support patients. Almost 30 million Americans suffer from rare diseases, so the summaries address an important healthcare problem. This year we crossed several new milestones.

1. We hit the mark of updating fifty NORD summaries!

2. Grad student Joe Farris with outreach coordinator Nurse Barb Calhoun developed the *first* new summary on Shprintzen Goldberg Syndrome for the NORD database. <https://rarediseases.org/nord-publishes-new-rare-disease-report-shprintzen-goldberg-syndrome-sgs/>



This was in partnership with a local family whose daughter (Hope) was the first to be diagnosed with this disease.

3. We contributed to the movies! NORD recently informed us that there's a new movie called "Wonder" starring Julia Roberts that tells the story of a child with a rare disorder affecting his appearance (Treacher Collins syndrome).

It's resulted in media links to NORD's report on that topic, which was updated with the help of Notre Dame NORD Intern, Joseph Lee (ND '13)!

[Joseph Lee – Treacher Collins Syndrome](#)



Example of how NORD's Rare Disease Database may be shared as a media resource: <http://www.refinery29.com/2017/11/173462/auggie-from-wonder-disease-treacher-collins-syndrome>

Rare Cancers and Rare Neurological Disorders.

Excellence in Graduate Education and Research.

CRND 'Catalyst' Program 2016-2017 Progress Report.

CRND Catalyst Projects are directed toward critical treatment targets or processes of drug resistance in cell, tissue, organismal and human systems. Four graduate students supported by the ***Boler family Endowment for Excellence***, made excellent progress in their Ph.D. thesis work. Well done!



Josh Mason

Rare Inflammatory Breast Cancer. Josh Mason (from the Schafer lab) studied inflammatory breast cancer (IBC), a rare, aggressive and difficult-to-treat breast cancer. Josh found that changes in the energy metabolism of IBC cells enabled their survival. Without this change, IBC cells can be killed by conventional drugs. Josh presented his findings at the Keystone Symposium on Tumor Metabolism in Whistler, BC in March 2017. He also published two papers on metabolism and cancer during this period. *Mason et al. Trends Cancer. 2017 Jul;3(7):475-481. Mason and Schafer Small GTPases. 2017 May 4:1-5. doi: 10.1080/21541248.2017.1310650.*



Ian Guldner

Rare Metastases to the Brain. Ian Guldner (from the Zhang lab) studied rare metastasis of cancer to the brain. He found that reducing immune cells in the brain, resulted in increasing brain metastatic burden. Based on these findings he used an FDA-approved drug that changed immune function to show it reduced metastatic cancer in the brain in a mouse model. Ian presented his findings at several meetings including American Association for Cancer Research Annual Meeting 2017 and three additional symposia at Notre Dame. A publication is planned. The research also led to the submission of a grant application to the National Cancer

Institute at the National Institutes of Health.



Ye Zhang

Rare Invasive Melanoma. Ye Zhang (D'Souza-Schorey lab) studies molecular properties of rare melanoma. Melanoma is a cancer of skin cells that contain the colored pigment melanin, making them easy to detect. But less than 5% lack pigment, and these rare melanomas are misdiagnosed as other conditions of the skin and very difficult to treat. Yi has identified molecular signatures that can be used in detection. In addition, when these molecular properties are removed (by genetic manipulation), they no longer spread on the skin, suggesting they also present targets for therapy. Presented at the ISEV (International Society for Extracellular Vesicles), Toronto, Canada 2017 and colloquia at Notre Dame. A manuscript is in preparation for journal submission.



Stefan Freed

Rare Neurological Disease, Non Ketotic Hyperglycinemia (NKH). Stefan Freed (Lee lab) develops multiparametric algorithms for predicting protein mis-folding applicable to a broad range of genetic disorders. He applied these tools to missense mutations of NKH, a rare and severe, neuro-metabolic disorder but with variable progression, the basis of which is poorly understood. Stefan developed automated analyses to evaluate established and new disease genes and compare them to changes in genes seen in healthy subjects. The findings enable predicting how severe the disease will be based on the patient's mutation (important information for clinicians and patient families) and guides selecting human mutations optimal for developing mouse models of NKH (needed to test therapies). Stefan presented his findings at Data Quality in the Era of Big Data (2016, IU Bloomington), and symposia at ND including the Shaheen 3-Minute Thesis, a University-wide competition where Stefan was a finalist and prize awardee (2017, Notre Dame). A manuscript is in preparation.

New Partnerships/Technologies/Strategies/Diseases

Ranedis-AMO. Ranedis Pharmaceuticals, who licensed CRND invention on brain-permeant formulations of HDACi shown to treat Niemann Pick Type C disease in preclinical mouse models (along with potential for other diseases DOI: [10.1126/scitranslmed.aad9407](https://doi.org/10.1126/scitranslmed.aad9407)), will further advance development of the formulation RND-001 through a collaborative agreement with AMO Pharma Ltd, as reported by Genetic Engineering and Biotechnology (GEN) News. <https://www.genengnews.com/gen-news-highlights/ranedis-and-amo-partner-to-develop-hdac-inhibitor-for-rare-genetic-diseases/81254020>

Investment in Genetic Methodologies. CRISPR-Cas9 is a technology adapted from a genome editing system of bacteria that has promise for genetically rectifying mutations that cause a broad range of rare diseases. For precision, speed, and potential for future optimization it is the lead genome-editing technology that has now been employed by CRND researchers to edit cellular systems and mice with mutations that cause Non Ketotic Hyperglycinemia (NKH), to develop treatments for the disease.

Collaborative Rare and Neglected Disease Teams Powered by Postdoctoral Research. A major challenge in many rare diseases, is that researchers who work on a given disease are located in widely separated institutions (indeed often, across countries). This makes it difficult to efficiently synergize different and complex strategies needed to accelerate to new discoveries and treatments. To counter, CRND has long utilized cross disciplinary training of graduate students. This year CRND initiated a platform for multi-lab collaborative work at the postdoctoral level (a more advanced stage of research training).

Intellectual Disabilities/Autism Spectrum Disorders/Pain. The unmet medical need provides the compelling drive to undertake research that leads to cures for a rare disease. But research in a rare disease can also provide potential for understanding and treating more prevalent disorders. The best example of this is the key discovery of statins that was made during a study a rare childhood disorder of hypercholesterolemia (with death due to heart attacks at ages as young as 5). Yet statins are widely used for lowering cholesterol in adults to reduce heart attacks and stroke. In contrast we have limited drugs for other complex but pressing conditions such as intellectual disabilities, autism spectrum disorders and chronic pain. These are major societal challenges, where CRND researchers have initiated work to additionally attempt extending the power of rare disease research to provide solutions.

Resistant Infections

Rare disease and mycobacterial infection



Cystic Fibrosis (CF), is one of the most prevalent of rare disorders. CF is a genetic disease that causes drying and thickening of the mucus along the nose, mouth, sinuses, throat, trachea, bronchial tubes, and lungs. These changes in the mucus make it difficult to efficiently clear germs, greatly increasing the likelihood of infections in the lung. One of the most common infections seen in CF patients and one of the hardest to treat is non-tuberculous mycobacteria (NTM) which colonize/infects approximately 13% of US CF patients. The biggest impact of an NTM infection in CF patients is decreased lung function and it is a significant cause of disease and death. Moreover, the prevalence of NTMs seems to be increasing. Clearly, new therapies for NTMs are needed.

Dr. Jeff Schorey (Department Biological Sciences) is an expert in mycobacterial infections. His laboratory studies both tuberculosis and NTMs. His work focuses on how these mycobacteria cause disease and the development of new therapeutics to combat mycobacterial infections including the evaluation of antibiotics. Recent studies, in collaboration with Marv Miller in the Department of Chemistry and Biochemistry and Yong Cheng in the Department of Biological Sciences, have examined a potent new class of anti-mycobacterial agents for their potential to treat NTM infections in CF patients. These compounds show excellent killing of a number of NTMs isolated from CF patients as well as from patients with other underlying lung diseases. The antibiotics also show activity in mouse infection models. Based on these initial studies, Dr. Schorey was recently awarded a grant from the Cystic Fibrosis Foundation to further develop and evaluate the efficacy of these antibiotics in a mouse model of CF.

Congratulations to Dr. Schorey!

Public Health in Bangladesh

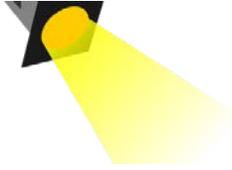
Public Health has markedly improved in Bangladesh over the last 30 years. One area of progress has been malaria control. There were 80,000 positive infections in 2008 compared to 27,237 in 2016. Over 90% of these cases occur in the Chittagong Hill Tracts (CHTs), at the border with neighboring Myanmar. Unfortunately, parasites in Myanmar are resistant to artemisinin drugs, fast-acting, frontline antimalarials that have greatly contributed to the recent gains in reducing malaria burden (and for which we still have no replacement drugs).



Spread of artemisinin resistance from Cambodia: black arrow shows spread to Myanmar. Red circles show regions with resistant parasites, that border Bangladesh and India and where ~50% of parasites are artemisinin resistant.

The rapid spread of artemisinin resistance throughout South East Asia has raised concern that it will spread to the rest of the world. Indeed the presence of artemisinin-resistant parasites at the Myanmar-Bangladesh border, suggests Bangladesh is in the path of spread to the rest of the world (akin to the spread of chloroquine resistance from SE Asia nearly 30 years ago), with potentially devastating effects for Africa. In an initiative supported by the Keough School of Global Affairs, investigators from CRND and Eck Institutes, the Departments of Biological Sciences and Chemistry and Biochemistry will undertake studies in partnership with the International Center for Diarrheal Diseases Research Bangladesh (icddr, b) to detect, still rare (or low frequency) but persistent, drug resistant infections and their transmission, in order to prevent spread of resistance and guide intervention strategies for both local and worldwide malaria control.

Part of this effort will also be channeled to establish a leading Public Health Science course with the Notre Dame Initiative for Global Development (NDIGD) at Notre Dame University Bangladesh (NDUB) in the science of disease elimination/eradication. One major educational outcome will be to integrate education and community engagement in understanding the science of malaria elimination/eradication in Bangladesh.



Spotlight



**Allison Maddox Slabaugh,
Director, Academic Advancement
College of Science, Department of Development.**

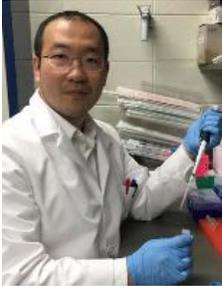
Allison has completed her first year as Director of Academic Advancement in the College of Science (but has been with Development at Notre Dame for 7 years). She helps Dean Mary Galvin with Development strategy. She's learned the burden of rare diseases, their unique challenges and the importance of expanding research and awareness. Research she says 'provides patient families with hope, letting them know they are not alone with their disease. 'When we are able to initiate research that is a huge step in their journey'. She finds working with patient families very rewarding and is grateful that ND has made a commitment to this research. Thank you Allison! Thanks also to the entire team of the College of Science and their leader Dean Mary Galvin!



**Corianne Kellems
Administrative Assistant,
Boler-Parseghian Center for Rare and Neglected Diseases**

Corianne joined the Center in March 2017. She is the central administrative and communications contact for the Center and supports the Director's position. Although her job has many facets, she especially enjoys connecting patients to resources and creating rare disease educational and awareness portals that include patients, researchers, clinicians and students. Her goal is to support vibrant communities that work together but also streamline CRND communication functions. As a first step she re-designed the Center website transforming it from 'a live blog' to a well-developed structure that supports our growth and diversity. She is trained in patient privacy and HIPAA (Health Insurance Portability and Accountability Act). She reminds 'Make sure you join us for Rare Disease Day 2018!'

Awards – Congratulations!



Dr. Xin Lu arrives and wins 2017 Indiana CTSI Young Investigator Award Dr. Xin Lu, the John M. and Mary Jo Boler Assistant Professor of Biological Sciences, in the Boler-Parseghian Center for Rare and Neglected Diseases arrived in January 2017. In less than a year he has set up a dynamic laboratory having rapidly recruited active graduate students and fellows reaching a lab size of 6!

In May 2017 he was awarded a Young Investigator Award from the Indiana Clinical and Translational Sciences Institute (ICTSI). Congratulations! Dr. Lu is a leading expert in developing mouse models that can mimic rare cancers caused by human mutations, in which to test new therapies. The award is to better target drugs to cancerous tissue and reduce harm to healthy tissue.

RareND Team builds ZEBRA, award-winning app in Notre Dame App Challenge and goal for Precision Medicine.

Medical Interns are counseled "when you hear hoof beats, think of horses not zebras", but 'Precision Medicine' seeks the zebra in all of us, leaving behind the

'average' patient to focus on individual differences and thereby understand a rare patient's disease and how to treat it. Congratulations to Anna Volk, Will Langbo, Kim Trochuck, and Katherine Inskeep of the RareND club for an important advocacy tool for patient-centered medicine and winning 3rd place in the first ND App Challenge!



2017 Ganey Award for Community-based research. Professor Kasturi Haldar, Nurse Calhoun, ND students in partnership with Beacon Pediatrics, Michiana

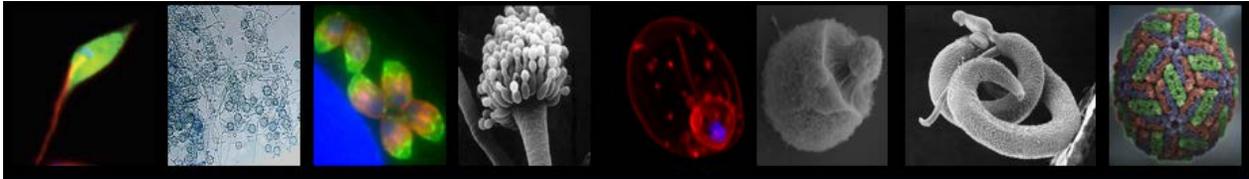


Health Information Network, received the 2017 Rodney

F. Ganey, Ph.D., Community-Based Research Award for a project that has helped improve rare disease recognition and treatment in within South Bend and surrounding area.

Conferences. CRND hosted its two major annual conferences.

The 6th Annual Midwest Neglected Infectious Diseases (MNID) Meeting 2017 was held on Aug 27-28th 2017, in partnership with six other Universities throughout the Midwest. It featured forefront research focused on the pathogenesis of fungal and parasitic diseases and was attended by scientists from Wisconsin, Illinois, Indiana, Michigan, Ohio, Iowa, and Missouri.



The next MNID meeting is scheduled for August 2018. Stay tuned for dates and detailed agenda.

CRND's 2017 Rare Disease Day Research Conference Feb 3-4, 2017 was a fantastic celebration with students, faculty, patients, researchers and clinicians. One young patient said it was better than going to Disney Land! The hockey was inspiring with ND coming from behind and then holding their own. A true rare disease inspiration! We are also deeply grateful for the assistance of Allen Utterback, Dr. Malgorzata Dobrowolska-Furdyna and Allison Maddox Slabaugh from the Dean's office, College of Science. Look forward to 2018.



CRND and you

We are not offering a quiz (this year) although we remain 'nerds' of strong foundations of facts and therapeutic discovery in rare and neglected diseases!

We invite you to participate in Rare Disease Day conference next year Feb 9-10, 2018 taking in dinner and ND-Ohio State Hockey Game on Feb 9, and keynotes by Nicole Boyce, a leader in Rare Disease Advocacy and Rob Long, a former all American punter who beat an aggressive rare cancer!

Thank you for your tremendous support in 2017. With your financial assistance we are able to undertake discovery and translational research to advance new therapies as well as support rare disease patient advocacy. We look forward to our journey with you in 2018.

We are humbled by your trust and faith in research and education at Notre Dame that can transform a child's life and are always grateful for your donation.

For NKH donations: [Click here](#)

For CRND Donations: [Click here](#)

Or mail check to CRND, 107 Galvin Life Sciences, Notre Dame, IN 46556

Rare Disease Day Symposium February 9th & 10th, 2018

BOLER-PARSEGHIAN
Center for Rare & Neglected Diseases



UNIVERSITY OF
NOTRE DAME
College of Science

Rare Disease Day®

February 9th - 10th

Jordan Hall - 9am-5pm

**Research Symposium / Exhibitor Booths / Posters
Rare Disease Patient and Research Leaders**

Key Note Speakers

Rob Long, Director Uplifting Athletes

Nicole Boice, Founder & CEO Global Genes



**ND vs Ohio State Hockey Game
Chuck a Puck: 7:30pm-10:30pm**

**To participate in exhibit booths / poster sessions,
please email: ckellems@nd.edu**

Event is FREE, but you must RSVP
by January 15th to ckellems@nd.edu
For more information, visit: <https://crnd.nd.edu/>