CRND Hosts Public Forum on World Rare Disease Day 2013

On Saturday, February 23rd we held our annual Rare Disease Day event. This year we joined patient families, faculty, students and alumni together to celebrate Rare Disease Day in order to raise awareness and increase advocacy for rare diseases. We celebrated a little early so that the community in our regional network could also attend. There were over eighty participants. We hosted four different discussion panels focusing on rare disease research, ethical dilemmas within rare disease work, patient family insights, and student outreach through our Rare Health Exchange (RHE) program, www.rarehealthexchange.org.
The Research panel was led by Notre Dame faculty Dr. Shaun Lee, a biologist who received the prestigious NIH Innovator Award in 2011. He collaborates with medicinal chemist Prof. Tony Serianni to find drugs that improve functions of poorly active proteins caused by genetic mutations that result in rare diseases. These drugs are called ‘chaperones’: they are important in treating many rare diseases. Other panelists Dr. Krastyu Ugrinov (post doc), Julie Chaney (graduate student) and Sue Yi (undergrad, class of 2013) also study chaperones and rare diseases at ND.

The Ethics in Rare Diseases panel was led by Dr. Carrie Quinn (ND alum ‘96 and pediatrician at Mt Sinai Hospital NY). Dr. Quinn, Brianna McSorely (ND alum, ’12) and current ND undergraduates, Lee Haruno (’13), Theresa Lai (’13), and Zach Weber (’13) began by presenting accepted codes of ethical conduct in medicine and biomedical research and then went on to discuss the ethics of disclosing medical information to children with rare diseases, new technologies that transform organ and tissue donation, and off label drugs that cannot be covered by Medicaid.

The Families panel was led by ND student Daniel Balcarcel who himself suffered transient episodes of Twin-to-Twin Transfusion Syndrome, a rare disease. The McLaughlin Family (ND alum) shared their story of BPES (Blepharophimosis, ptosis, and epicanthus inversus syndrome), an eyelid malformation that can severely affect vision and fertility. Koujaian and White family narratives of NPC (Niemann Pick Type C disease, a neurodegenerative lipid storage disorder) reinforced the challenges in obtaining diagnosis and treatment in rare diseases because of lack of information and awareness.

Forging Family Ties

Thanks to all for their participation in the 2013 ND World Rare Disease Day celebration
The Notre Dame Rare Disease Fight Song:

Rally students of Notre Dame,
Fight rare disease, and sound her fame
Raise her Gold and Blue,
And cheer with voices true,
In the fight against rare disease,
Fight rare disease in every game
Strong of heart and true to Notre Dame.
We will ne'er forget her
And we'll cheer her ever,
In the fight against rare disease.

Chorus:
Fight rare disease at Old Notre Dame
Ending their neglect and cheering her name,
Send the volley cheer on high,
Shake all seven thousand down from the sky,
What though the odds be great or small
Old Notre Dame will win over all,
While her loyal students march
In the fight against rare disease.

See the Glee Club Inauguration of the ND Rare Disease Fight Song in 2012:
http://vimeo.com/37980822

Marisa Truong program coordinator (and ND alum, ‘11) and Steve Kraska (Computer Science, ‘13), led discussion on a custom-designed Rare Disease database at ND established by collaborating with family foundations and the Michiana Health Information Network (MHIN; Hannah King and Scott Kidder). Its goal is to be a patient-centric database that can be searched within and across rare diseases to assist patients and researchers.

Two additional highlights were (i) lunch, where everyone worked hard at the RHE sponsored rare disease quiz and (ii) the finale of the Rare Disease Fight song, led by Dr. Lee and members of the ND Band!

A Rare Disease Quiz for Lunch
Building Bridges: Science, Engineering & Beyond

Rare Health Exchange is a collaborative of pre-medical students trained to assess rare disease patient medical records and assist in the development of natural histories (http://rarehealthexchange.org). The Rare Health Exchange Database was built through interdisciplinary collaboration between CRND and iCeNSA as well as the National Niemann Pick Disease Foundation (special thanks to Karen Quandt, Adam Ward and Jenn White) and the Michiana Health Information Network (MHIN; special thanks to Hannah King and Scott Kidder). It was supported by CRND and an Edison Innovation Award by Dean Gregory Crawford in the College of Science.

_The goal is a patient-centric database that can be searched within and across rare diseases to assist patient families and researchers._

Marisa Truong, Program Coordinator, CRND (class of ‘11) and Steve Kraska (Computer Science, ‘13), established the custom-designed database for RHE. RHE students have entered symptoms and their severity for two rare diseases Niemann Pick Type C and Congenital Hypothyroidism.

Through BIOS40450 (Developing Health Networks in Rare and Neglected Diseases, taught by Professor Haldar) ND students become trained to be active RHE participants. As Program Coordinator, Marisa Truong connects the students with rare disease organizations and patient families (http://rarehealthexchange.org/patient-family-interaction-training.html). She also helps students organize often hundreds of pages of patient data, condense them into 2-3 page disease summaries which are helpful to patients. Students then enter symptom and disease severity data into the RHE database. Marisa ensures that patient privacy is protected and has worked closely with Steve Kraska on development of the RHE database – an inspired collaboration between Science and Engineering. Next year, she looks ahead to another phase of inspired training at the prestigious nursing program at DePaul University, Chicago.

Steve Kraska (ND ‘13) a senior in computer science, who works with Prof. Chawla, designed the database to easily add on any number of diseases and their associated symptoms. This is an important consideration for a database for potentially thousands of diseases. Right after graduation he will be working as a database and data mining consultant for West Monroe Partners in Chicago. He says the “RHE Data base experience has been crucial in my preparation for the consulting world because I have had an
In the fight against rare diseases, a major hurdle is collecting reliable information about a condition that may affect only a few patients. The next challenge is to do it again for the roughly 7,000 diseases designated by the National Institutes of Health as rare.

Dr. Nitesh Chawla is a Notre Dame researcher (Computer Science) who is at the forefront of Big Data research. With health records moving from being on paper to being electronic, Dr. Chawla is developing innovative algorithms with a focus on personalized healthcare, disease prediction, and patient centered outcomes, for both chronic and rare diseases.

Dr. Chawla’s involvement with the RHE database has been essential to the project’s success. “It is a project with a great potential to bring data and networks to make a difference to the patients and families with rare diseases”, noted Dr. Chawla. The goal is to “apply my research program in personalized healthcare for making a difference, to enhance the speed and opportunity of prognosis, to enable patients and physicians to come together for diagnosis and prognosis at the earliest opportunity. It also provides a platform for families to connect with each other and share experiences.”
Dr. Chawla started his tenure-track career at Notre Dame in 2007; he was tenured and promoted in 2011 and recognized with the Frank Freimann Collegiate Chair in 2012. He is the director of the Notre Dame Interdisciplinary Center for Network Science and Applications (iCeNSA). He serves on numerous prestigious editorial boards and is the recipient of multiple awards for research and teaching innovation, including most recently the IBM Watson Faculty Award. He is also the founder of Aunalytics, Inc, a start-up focused on analytics and housed at Notre Dame’s Innovation Park.

When he’s not making his way through ‘Big Data’, Dr. Chawla enjoys spending time with his family, voraciously reading books, and experimenting with food.

His main regret is that his 2002 Ph.D. thesis refers to ‘massive’ not ‘big’ data!

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### Center Highlights

#### “Rare ND” Club Established

The newest student club is here! “RareND” is now established as a club through CRND ([http://rarehealthexchange.org/rare-neglected-disease-student-clubs.html](http://rarehealthexchange.org/rare-neglected-disease-student-clubs.html)). The purpose is to develop a student club engaged in discussions, contests and fundraisers to increase awareness of rare diseases. The founding president is Joey Kim (ND ‘13).

#### Sue Yi, the McDonald Award Recipient

Notre Dame undergraduate Sue Yi received the 2013 “Mr. & Mrs. Frank McDonald Undergraduate Research Award” from the Department of Biological Sciences. Sue’s outstanding work on the rare disease Niemann-Pick Type C in the Haldar lab has resulted in a co-authored publication, whose content was a plenary lecture by Dr. Haldar at the March 2013 ‘Brains for Brain’ Meeting in Frankfurt Germany. Please join CRND in congratulating Sue Yi for her accomplishments.

#### New Administrative Assistant for CRND

In March 2013, Paul Lingle joined the CRND team as the new administrative assistant. Paul hopes to “take CRND to the next level, providing quality services to all our associates and the rare disease patient community”.

Paul brings 10 years of office management at Saint Joseph Regional Medical Center. Prior to that he was in the video/graphics editing industry. Please join us in welcoming Paul.
NORD Editorial Internship for ND Undergraduates

The idea was to develop an educational and service program to help the National Organization for Rare Diseases (NORD) continuously update its vast library of rare disease summaries. These summaries are an invaluable resource for families afflicted with a rare disease, where they can learn about its genetic basis and progression, physicians and centers that treat and study the disease, as well 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.

*Thank you to the Fall 2012 Class and Congratulations on Graduating!*

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**Test your Rare Disease Knowledge Against Theirs.**

1. Even though Jervell & Lange-Nielsen syndrome equally affects both males and females, the disease: A. is more aggressive in males. B. presents earlier in females. C. has less life-threatening events in females. D. None of the above.

2. Individuals with Hyperekplexia react to unexpected stimuli: A. Slowly. B. Excessively. C. Do not react. D. They react even without stimuli.

3. Cardiofaciocutaneous syndrome symptoms include all of the following except: A. Poor growth and dry skin. B. Heart abnormalities. C. Hearing Loss. D. Cognitive delays.

*For Answers, Use the Links to Student Summaries (right panel) or go to:*

http://rarehealthexchange.org/news/?p=331

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**NORD Editorial Interns**

- **Theresa Lai** – Jervell and Lange-Nielsen:
  

- **Sarah Fagan** – Cardiofaciocutaneous Syndrome
  

- **Michelle Yanik** – Incontinentia Pigmenti
  

- **Zachary Weber** – Thrombocytopenia Absent Radius
  

- **Lee Haruno** – Hyperekplexia
  

- **David Brouch** – Fanconi Anemia
  

- **Joey Kim (Founder)** – Brugada Syndrome
  

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**Soon to be published!**

- Daniel Balcarcel
  
  Pallister-Hall

- Joseph Lee
  
  Treacher-Collins
Additional Updates on targets and therapies:
CRND’s Focus on Treatments for Neglected Infectious Diseases

World Malaria Day

World Malaria Day, April 25, was a chance to shine a spotlight on the need for new drugs to eradicate malaria.

See the “Speaking of Medicine” Feature in PLoS BLOGS: http://blogs.plos.org/speakingofmedicine/2013/04/25/malaria-targets-and-drugs-for-all-stages/

CRND Releases New Video

A new video produced by CRND highlights the efforts of the researchers at the Center for Rare and Neglected Disease to eradicate malaria. The video shows the partnership between CRND, Eli Lilly & Co and the Medicines for Malaria Venture.

Watch the video at: http://vimeo.com/65411648

The 3rd Annual Midwest Neglected Infectious Diseases Meeting

August 23-24, 2013
Jordan Hall, Notre Dame

Dr. Aaron Mitchell
Carnegie Mellon University

Keynote Lecture: “Transcriptional Control of Candida Virulence”

Dr. Marvin Miller
University of Notre Dame

Keynote Lecture: “New Antibiotics to Address the Never Ending Microbial War”

for more information, visit: www.nd.edu/~crnd