The 2015 NORD Rare Diseases and Orphan Products Breakthrough Summit

The Snyder-Robinson Foundation was fortunate to receive a scholarship from the National Organization for Rare Disorders (NORD) to attend The Rare Diseases and Orphan Products Breakthrough Summit. The Summit featured over 20 speakers from the FDA, and included participation from over 80 patient organizations and the Pharma/Biotech industry's foremost experts in orphan product innovation, investment and commercialization.

The Summit contained innovative content and convened the top leaders from the FDA, NIH, Industry, Patient Groups, Payers and Research Institutions to address the progress of rare disease diagnosis, genomics, drug development, patient engagement, product approvals, FDA oversight and market accessibility to orphan products.

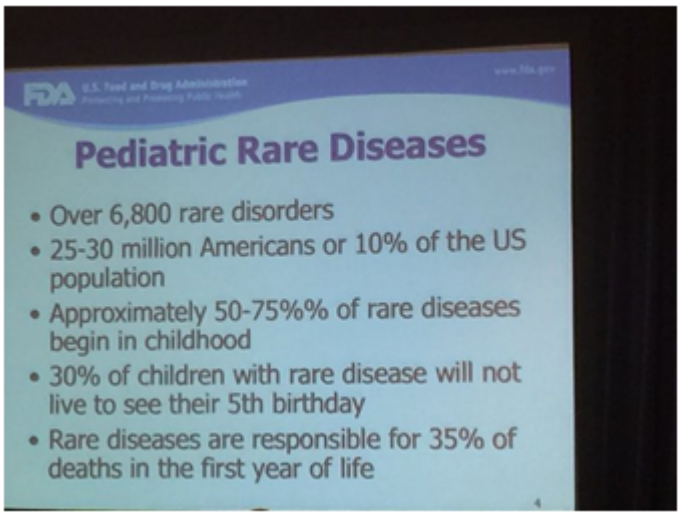
An overarching theme of the Summit was the importance of the patient voice. Lynne Yao, M.D., the FDA Acting Director of the Division of Pediatric and Maternal Health, said, "through the patient voice, we have learned so much more." Another theme that ran throughout the conference was the importance of patient advocacy organizations. An enormous change has occurred with

rare diseases because they can now make connections and unite globally, and can get together in a way that was not possible before. This connection leads to huge differences in patient involvement in rare disease research. In addition, Mr. Sachdev, of VertexPharma, stressed how important it is for rare disease communities to have “robust registries” that include natural history studies and the burden of the disease. Janet Woodcock, FDA Director, observed that “the future has never been brighter for rare diseases.”

“Promoting Earlier Diagnosis Through Physician Education and Awareness” was a very interesting panel with a lot of discussion into topics relevant to the SRF and Snyder-Robinson Syndrome. The Panel included Robert Saul, M.D., the co-Director of the American Academy of Pediatrics “Genetics in Primary Care”, Richard Peters, of Genzyme, Mary Dunkle, Vice President, NORD, who is heading up NORD’s new Educational Initiatives department, to expand outreach to current and future medical professionals, Steve Kaminsky, the Chief Science Officer of International Rett Syndrome Association, Sophia Walker, a medical student at the University of Connecticut, who is involved in rare diseases advocacy and is serving as a medical student liaison at NORD.

The panel members stressed the importance of earlier diagnosis through physician education. Because there are over 6,000 rare diseases, physicians are presented with a huge challenge. The goal is to make diagnosis faster and then to work with the FDA to get treatments. Mr. Peters explained that industry has an important role to play in funding resources to raise awareness of diseases. Mr. Saul stated that the challenge is how to ingrain genetics into new physicians, and to teach them that even if you are not able to treat the disease, you can always help to treat the patient. Mr. Saul observed that genetics is

the fabric of primary care, not just for rare and unusual conditions. Mr. Kaminsky explained how his organization is trying to empower families to receive the care they need across the country, and to connect with their communities. He also advised people living with rare diseases to build a medical record to take with you to the ER that explains your condition, along with Doctor contacts.



As a NORD member organization, The Snyder-Robinson Foundation was invited to spend one of the conference days learning about Patient-Centered Outcomes Research (PCOR), and to attend a special training for Rare-Disease Patient Advocates. This portion of the Summit was cosponsored by NORD and the University of Maryland, School of Pharmacy, Baltimore. The training was aimed to help advocates understand how to use patient engagement methods to strengthen PCOR and how their organizations might take part in some of them. It was also designed to develop communication and networking skills and strategies to promote meaningful interactions with PCOR stakeholders to promote partnership and to help advocates be able to learn the key features of successful PCOR proposals and projects.

This training was invaluable, as much of the funding of rare disease research now comes from PCOR, which was set up by The Affordable Care Act in 2010. I learned as much as I could in a short amount of time, and made contacts that I can utilize when questions arise. I learned about the Roadmap to Patient-

Focused Outcome Measurement in Clinical Trials, which is on the FDA website. Every patient group must go through this process ahead of time, in development, before they do anything else. That way, when a clinical trial of a therapy is approaching, road blocks will not suddenly appear. As a result of the Patient Advocates Training, the Snyder-Robinson Foundation is now much better positioned to apply for a PCORI Pipeline Proposal Award.

One of the most relevant presentations in this portion of the Summit was entitled “The Natural History of a Rare Disease.” This presentation focused on how important it is to have the natural history of a disease and to treat the information as a leverageable asset of the patient advocacy group. Further, the presentation explained how data sources such as the registry, surveys and focus groups can be utilized to support PCOR partnerships. The Natural History of a disease helps to know if the treatment is helping the patient. It also helps scientists learn generally about the human body and how the body works. The term Patient Focused Drug Development means that we need to understand what the benefits of a drug are from the patient’s point of view. This is very different from prior models that utilized a clinician’s interpretation of a drug’s benefit to his/her patient.

During the Summit, Mr. Sheldon Schuster, a member of the NORD Board of Directors, stated that “the most powerful force in the universe is the parent of a child with a rare disease.” As a parent-led volunteer advocacy organization, the Snyder-Robinson Foundation couldn’t agree more. Our natural history study is underway, and we look forward to the value it will bring for SRS and the Foundation!

– Katia Luedtke