RARE DISEASES NEWSLETTER

Volume 12, February 2019

WELCOME TO THE TWELFTH EDITION OF THE RARE DISEASES NEWSLETTER.

Welcome to the twelfth edition of the rare disease newsletter. This quarterly publication strives to keep you up to date on PRA's Center for Rare Diseases, our achievements, and new initiatives.

Interested in learning more about our Center and/or how we can support you? Email us – we take requests! To learn more or to obtain support for a specific clinical study or project, please contact us here. You can reach the entire team by emailing us at PRACenterForRareDiseases@prahs.com.

EDITION HIGHLIGHTS

RARE DISEASE DAY 2019

This year we are excited to be hosting PRA's first annual World Rare Disease Day event on February 28th, 2019. The event will be held at the PRA office in Blue Bell, PA from 11 AM – 3 PM. Speakers from rare disease patient communities will discuss their journeys and how rare diseases have impacted their daily lives.

We hope that this event will drive awareness of the over 7,000 rare diseases and the impact they have on 1 out of every 10 people. Click here to review the agenda and register your attendance.

THE RARE DISEASE TEAM WARMLY WELCOMES JESSICA WESSEL

JESSICA WESSEL
Clinical Research & Opthalmology
Rare Diseases

Holding Certifications in Clinical Research and Ophthalmology, Jessica has committed her career to the acceleration and development of both new and existing treatments for Ophthalmic indications. With over 5 years of experience and expertise in operational feasibility based on previous cultured academia experience in an array of indications, from anterior to posterior segments.

Prior to joining PRA, Jessica worked as Senior Clinical Research Coordinator and Compassionate Use Liaison at the Cincinnati Eye Institute where she managed over 20 commercially sponsored, phase II-IV, interventional, IND and IDE trials in addition to the submissions and approvals of 21 Compassionate Use Device Exemptions. Jessica has assisted with the writings and considerations for several Investigator Initiated Trials in addition to co-creating and presenting various presentations on the topic of Compassionate Use.

Jessica is a native Cincinnatian where she presently serves as Local Chapter President for the Association of Clinical Research Professionals (ACRP) in addition to other membership roles for assorted organizations within her community. In her new role with PRA, Jessica will serve as a therapeutic expert and will help lead continued growth of our Ophthalmology offerings.

RARE DISEASE INDICATIONS - HIGH ACTIVITY

- Pompe Disease
- Achondroplasia
- Charcot-Marie-Tooth Disease
- Rett Syndrome
- Hemophilia
- Pemphigus
- Retinitis Pigmentosa
- Sickle Cell Disease
- Metachromatic Leukodystrophy
- Duchenne Muscular Dystrophy
- Limb Girdle Muscular Dystrophy
- Amyotrophic Lateral Sclerosis
- Neuromyelitis Optica
- Friedreich Ataxia

DISEASE SPOTLIGHT:

CHARCOT-MARIE-TOOTH DISEASE (CMT)

Peripheral nerves are wires that carry the messages from the brain and spinal cord to the muscles. When either the core (axon) or insulation (myelin) of these peripheral nerves are damaged, this results in weakness, numbness and pain. This condition, called peripheral neuropathy, is most commonly seen secondary to disorders like diabetes mellitus. When peripheral neuropathy occurs due to genetic mutations, it is called Charcot-Marie-Tooth disease, or CMT.

Named after the physicians who initially described the condition in 1886 (Jean-Martin Charcot, Pierre Marie, and Howard Henry Tooth), CMT most commonly presents in childhood and early adolescence with weakness of the distal muscles in the feet and hands, making it difficult for patients to walk and perform fine motor tasks. Over time, the weakness progresses to the proximal legs and arms. Most patients require ambulatory aids such as ankle-foot orthoses to walk, and some progress to requiring canes, walkers and wheelchairs for mobility. CMT is a rare, but the most commonly inherited peripheral nerve disorder. CMT affects 1 in 2500 people in the US and roughly 2.8 million people worldwide. The first mutation causing CMT was identified in a single gene in 1991; at present, we have identified over 300 mutations in 80+ genes (and counting) that result in CMT. Thus, CMT is not a single entity, but rather a spectrum of peripheral neuropathies that result from genetic mutations. CMT can occur as a brand new (de novo) mutation or can be inherited in several ways: autosomal dominant (through a faulty gene contributed by either parent); autosomal recessive (through a gene contributed by each parent); or X-linked (through a gene on the X chromosome contributed by either parent in girls, or by the mother in boys). CMT is classified broadly into CMT1 (myelin damage) and CMT2 (axon damage); the term Dejerine-Sottas syndrome (replacing the previously used CMT3) is used to describe severely affected infants with CMT; CMT4 is used for autosomal recessive CMT, and CMT1X stands as its own category. With the discovery of the causal genetic mutations, letters have been added to indicate the gene. For example, peripheral myelin protein gene (PMP22) duplication causes CMT1A, the most common type of CMT; mutations in the mitofusin 2 gene (MFN2) cause CMT2A, etc.

Although CMT is not often fatal, it is certainly life altering - CMT is progressive and currently incurable. As our understanding of the diseases has increased over recent years, the Center for Rare Diseases is excited to see new therapies in development for these patients. Also, the CMT patient advocacy community is doing some incredible things! WATCH HERE as Allison Moore, Founder and Chief Executive Officer, Hereditary Neuropathy Foundation discusses the innovative and game changing things that patient advocates are doing in CMT!

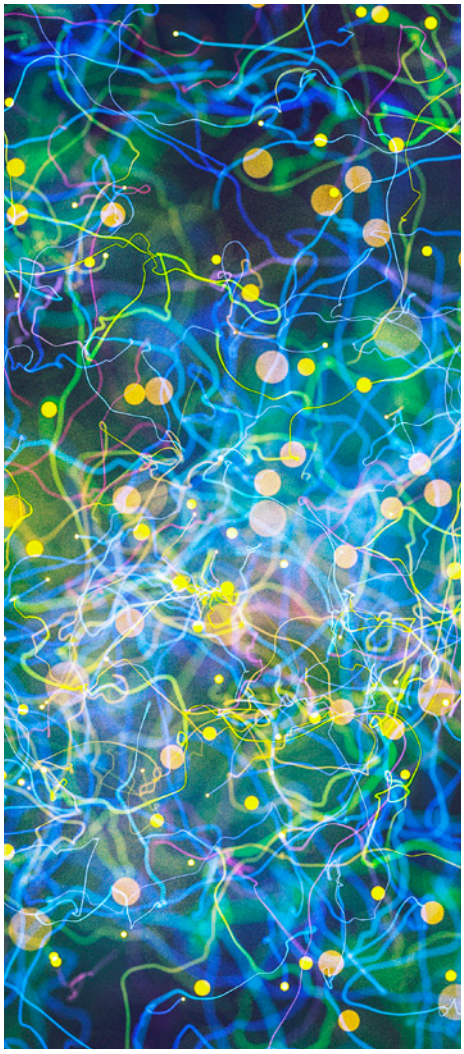
NEWLY AWARDED STUDIES:

- Spinal Muscular Atrophy – Long Term Follow Up
- Phenylketonuria (PKU) – Phase I
- Recessive Dystrophic Epidermolysis Bullosa (RDEB) – Phase I/II
- ATTR-Cardiomyopathy – Phase III
- Primary Sclerosing Cholangitis – Phase III
- Pseudoxanthoma elasticum – Phase IV

WANT TO LEARN MORE?

Check out the links below to some of the incredible foundations supporting CMT!

- CMT Research Foundation
- Cure CMT4J/Talia Duff Foundation
- Charcot-Marie-Tooth Association (CMTA)
- Heredity Neuropathy Foundation
- Muscular Dystrophy Association



MEETING HIGHLIGHTS

MAGI | SAN DIEGO, CA | OCTOBER 2018

Derek, Scott, and Amy presented at the Model Agreements & Guidelines International (MAGI) West conference in San Diego, CA on October 21-24, 2018. MAGI is an organization focused on providing best-in-class resources for clinical sites. In addition, MAGI supports regulatory, finance, and operational teams by standardizing document templates that expedite the clinical trial start-up process.

Amy presented on Sunday in a workshop entitled "Clinical Project Management: A Skills-Based Workshop". She, along with her co-presenters, provided project management best practices for aspiring or current project managers in the industry.

Scott presented "Bringing the Study to the Patient" where he discussed the industry trend of decentralized trials. He also participated in panels entitled "Rare Disease Clinical Research: A Panel Discussion" and "Stump the Experts".

Scott Schliebner on stage with fellow panelists during the Clinical Development session of World Orphan Drug Congress (November 2018).

Derek represented PRA in a panel focused on patients, entitled "Patient-Centric Clinical Trials: A Panel Discussion". The panel focused on different vendors used in decentralized trials, such as home nursing care and digital devices, and the common challenges that accompany these solutions. Click here to learn more!

WORLD ORPHAN DRUG CONGRESS

Derek, Scott, and Amy were joined by colleagues from Medical Informatics, Marketing, Business Development, and Real World Solutions at the World Orphan Drug Congress in Barcelona, Spain on November 6-8, 2018. This meeting is designed to foster collaboration, patient advocacy, enhance strategy, and partnership in the global rare disease global research community.

One of the many benefits of this international gathering is the forum for patient advocacy groups to helpfully shape drug development efforts and collaborate by sharing best practices. Scott was selected to present "What Does a Patient-Focused Clinical Trial Really Look Like? Disrupting the Current Clinical Trial Paradigm" in the clinical development track. Scott and Derek also co-hosted a roundtable discussion entitled "The Virtuality of Rare Disease Clinical Trials: Where Are We Headed?".

The PRA team joined clinical research colleagues and a group of clients on a guided tour of Casa Mila, the last modernist structure completed during architect Guidi's lifetime. Pictured, from Left: Amy Raymond (Center for Rare Diseases), Scott Schliebner (Center for Rare Diseases), Leyla Honar (Medical Informatics), Emma Watson (Marketing), Alex Malouvier (Real-World Solutions), and Derek Ansel (Center for Rare Diseases).

The PRA booth was a hub of activity, and additional conversations took place during the PRA-hosted private tour of Casa Mila (also known as La Pedrera). This was a fantastic opportunity to soak up some local culture at this UNESCO World Heritage Site with fellow clinical researchers from around the globe. We are grateful for the opportunity to make many new contacts within the rare disease community, connect with additional patients, and meet future drug development partners. Looking forward to continuing these conversations into 2019!

2018 FOURTH QUARTER HIGHLIGHTS

WHERE WE'VE BEEN

- Jana, Amy, and Scott attended and each presented at the International Society for CNS Clinical Trials and Methodology (ISCTM) Autumn Conference, 15-16 October, Marina Del Rey, CA
- Scott, Derek, and Amy attended and presented at MAGI West Clinical Research Conference, 21-24 October, San Diego, CA
- Sravan attended and presented at Early and Managed Access Programmes, 22-24 October 2018, London, UK
- Amy, Derek, and Scott attended the World Orphan Drug Congress, 06-08 November 2018, Barcelona, Spain
- Derek attended the American Society of Hematology Annual Meeting, 01-04 December 2018, San Diego, CA

One of two posters presented by the Center for Rare Diseases at the International Society for CNS Clinical Trials and Methodology (ISCTM) poster session in October 2018.

IN THE PIPELINE:

- Systemic Sclerosis – Phase I/II
- ATTR Amyloidosis – Phase I/II
- Gaucher Disease – Phase I/II
- Rett Syndrome – Phase I/II
- Molybdenum Cofactor Deficiency (MoCD) – Phase I/II
- Hemophilia – Phase I/II
- Achondroplasia – Phase II
- PKU – Phase II

WANT TO KNOW HOW CLINICAL RESEARCHERS ARE USING INNOVATION TO DRIVE SUCCESS IN CLINICAL DEVELOPMENT FOR RARE DISEASES?

Learn more in this article on Rare Disease Innovation in PharmaVOICE found here.

CONTACT INFO

If you need assistance with a rare disease study, have a personal interest in rare diseases, or would like more information, please contact us at PRACenterForRareDiseases@prahs.com or individually:

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