WELCOME TO THE SIXTH EDITION OF THE RARE DISEASES NEWSLETTER!
This quarterly publication will keep you up to date on PRA’s rare disease team, experience, achievements, and initiatives.

EDITION HIGHLIGHTS
01. World Orphan Drug Congress 2017
02. Disease Spotlight: Huntington’s Disease & Diseases of Protein Metabolism
03. Where We’ve Been & Where We’re Going

WORLD ORPHAN DRUG CONGRESS RECAP
In April, the PRA team attended the World Orphan Drug Congress (WODC) in Washington, DC. This 3-day conference provided a platform for different stakeholders from within the rare disease community, including patients, advocacy groups, pharma & biotech companies, and CROs, to learn about and discuss challenges that face rare disease patients and their caregivers.

PRA hosted 2 successful roundtable discussions at WODC:
2. Biosimilar Orphan Drug Development - preparing for an inevitable reality and a panel discussion on - “Is the orphan drug market ready to embrace the biosimilar landscape?”

Hazel Gorham also gave an excellent talk on biosimilar orphan drug development. In addition, PRA also hosted an extremely successful private cocktail reception at Stone’s Throw Restaurant, which generated several new connections and business opportunities with new rare disease clients, vendors, and partners.

The PRA brand was evident throughout the entire event, from the PRA motion graphic booth to hundreds of PRA’s canvas bags. Our team held 15 client meetings and generated several new business relationships within the rare disease space.

RARE DISEASE BOOTH AT WORLD ORPHAN DRUG CONFERENCE IN WASHINGTON, DC.
DISEASE SPOTLIGHT:

HUNTINGTON’S DISEASE AWARENESS MONTH

May is Huntington’s disease Awareness month and with the recent approval of the 2nd therapy in this indication, and the first in a decade, we thought it a great time to highlight this disease and the promising work taking place in this field.

Huntington’s disease (HD) is a fatal neurodegenerative disease, characterized by the progressive breakdown of nerve cells in the brain leading to uncontrolled movements and cognitive deterioration. HD is inherited in an autosomal dominant fashion, meaning there is a 50/50 chance a parent with HD will pass this disorder onto each child, and is not dependent on sex, race, or ethnicity. HD has been described as having symptoms of ALS, Parkinson’s, and Alzheimer’s all at once.

HD is most often diagnosed in a person’s prime adult years, between the ages of 30-50, although there are individuals who experience juvenile onset. There is no cure for HD and treatment through the stages of HD focuses on alleviating depression and therapy to maintain control over voluntary movements to care for oneself. In late stage HD total care is required and the ability to speak, chew, and swallow are lost although comprehension and awareness of one’s surrounding may still be intact. It is estimated that more than 30,000 individuals in the U.S. are currently living with HD. The Huntington’s Disease Society of America (HDSA) is a non-profit organization which has partnered with 41 Centers of Excellence throughout the United States to provide multidisciplinary care and research for individuals and families affected by the disease. During the last 5 years, PRA has conducted over 264 studies in neurology indications including Huntington’s disease (HD) and an additional 22 studies in a variety of movement disorders. Our significant corporate experience gives us an understanding of the specific nuances of running these studies including ensuring quality data, minimizing placebo response and leveraging our relationships with the multiple stakeholders such as Huntington’s Study Group, European Huntington’s Disease Network, patients, KOLs and caretakers.

In April of this year the FDA approved AUSTEDO™ (Teva Pharmaceuticals) which showed significant improvement in controlling the involuntary movements, termed chorea, experienced by nearly 90% of HD patients. The HD community is no doubt celebrating the achievement this May as the HDSA slogan reminds us all to continue to seek “Help for Today, Hope for Tomorrow.”

DISORDERS OF PROTEIN METABOLISM

Disorders of protein metabolism represent several distinct rare disorders all of which currently are areas of significant unmet medical need. To date few treatments exist beyond strict dietary restriction. These disorders either involve a block in a specific enzymatic pathway to break down protein into their component amino acids, or the inability to detoxify a product of a protein metabolism pathway such as ammonia. Dietary therapy is an essential component of treating these disorders and focuses on the restriction of protein intake and in most cases, especially pediatric, supplementation with a specialized metabolic medical formula to achieve nutrition requirements for supporting adequate growth and development. Despite treatment with diet and medical formulas, significant morbidity continues to exist. If left untreated these disorders result in severe neurological and cognitive impairment and may result in mental retardation, seizures, and life threatening elevations in ammonia which can result in profound brain damage and death from organ failure. In the last 30-40 years newborn screening programs have greatly improved the early identification of affected individuals however, screening programs are not uniform nor are they all-inclusive and many affected individuals and families are burdened by a prolonged diagnostic odyssey. Few treatments exist beyond strict dietary restriction with Phenylketonuria, Primary BH4 Deficiency, and Urea Cycle Disorders having collectively three approved medical treatments. Maple Syrup Urine Disease, a defect in the metabolism of Branched Chain Amino Acids, named originally for the sweet smelling urine of affected individuals, has no approved treatment and currently only one registered, single-site investigator initiated trial. PRA proudly supports the ongoing search for viable treatments to normalize the quality of life for these individuals and is conducting 5 trials in Phenylketonuria while actively engaging with biotechnology and pharmaceutical partners developing treatments for these underserved populations.

RARE DISEASE INDICATIONS WE ARE ACTIVELY PURSuing:

- Huntington’s disease
- Non-alcoholic Fatty Liver Disease
- Beta Thalassemia
- Sjogrens Syndrome
- Friedreich’s Ataxia
- GNE Myopathy
- Amyotrophic Lateral Sclerosis
- Primary immunodeficiencies
- Myasthenia Gravis
- Neurotrophic Keratitis
- Hunter Syndrome
- Retinitis Pigmentosa
- Adrenoleukodystrophy (ALD)
- Cystic Fibrosis
- Phenylketonuria
- Hemolytic Disease of the Fetus and Newborn
- Atypical Hemolytic Uremic Syndrome (aHUS)
- Enteric Hyperoxaluria
- X-Linked Hypoposphatemia
FIRST QUARTER HIGHLIGHTS

NEWLY AWARDED RARE DISEASE STUDIES:
- Primary Biliary Cirrhosis - Proof of Concept Trial
- Leber’s Hereditary Optic Neuropathy Gene Therapy Extension Study X2

WHERE WE’VE BEEN
- Scott and Sravan attended WODC in April (along with John Mann, Danielle Libero, Hazel Gorham, Jackie Zimmerman, Lisa Cilento, and Dawn Joseph)
- Lisa attended the SMA Patient-Focused Drug Development Meeting with the FDA in April
- Scott attended and presented at the ACRP/PharmaTimes Clinical Researcher of the Year event in April

WHERE WE’RE GOING

May 2017:
- Lisa at the 13th International Conference on Myasthenia Gravis and Related Disorders 15-17 May 2017
- Scott at UPenn/Global Genes Patient Advocacy Summit in Philadelphia, PA 19 May 2017
- Scott will present at Linking Leaders in Philadelphia, PA 23 May 2017
- Sarah will attend the Pediatric Academic Societies Meeting in May in San Francisco, helping to launch the Center for Pediatric Clinical Development

June 2017:
- Scott will lead a podcast/webinar for ACRP on 06 June 2017
- Scott and Lisa will attend and present at DIA in Chicago, IL in June

September 2017:
- Looking ahead to Global Genes Sept Tribute and Gala - PRA Title Sponsor

CONTACT INFO

If you need assistance with a rare disease study, have a particular personal interest in rare diseases, or would like more information, please contact us at:

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WILSON BRYAN, MD
Director,
Office of Tissues & Advanced Therapies at the FDA

Wilson Bryan, MD made the opening remarks in April at Patient Focused Drug Development Meeting for Spinal Atrophy regarding the mutual education process between patients and regulators that makes drug development most productive and effective.