PDSA Attends NIH Rare Disease Day 2012

PDSA joined other patient support groups on Leap Day, February 29th, as the National Institutes of Health (NIH) celebrated the fifth annual Rare Disease Day. The one-day gathering at NIH in Bethesda, MD was to recognize the various rare disease research activities supported by the NIH Office of Rare Diseases Research, the NIH Clinical Center, other NIH Institutes and Centers, the Food and Drug Administration (FDA) Office of Orphan Product Development, the National Organization for Rare Disorders (NORD), and the Genetic Alliance.

The Director of the Office of Rare Diseases Research, Dr. Stephen Groft, welcomed the attendees and gave an overview of research conducted by the Office of Rare Diseases Research at NIH. Next, Dr. Francis Collins, the Director of NIH, described how researchers might be able to 'leap forward' in this important area of research. He said there are 7,000 rare diseases and now over 4,000 have had their molecular underpinnings discovered. Dr. Collins said thanks to less expensive gene sequencing now the gap between what we know and what we can do about it has declined.

He added, "We need our 'wisdom' to be of benefit to patients with rare diseases and disorders, in some cases success comes with repurposing of some drugs developed for other purposes." Dr. Collins said NIH is working to break down barriers to researching new therapeutic treatments sooner. He said, "These are challenging but exciting times." A new resource was announced to help caregivers gain access to the NIH Genetic Testing Registry for information about genetic tests. The Web site is: http://www.ncbi.nlm.nih.gov/gtr

Presentations were given by 16 scientists from various NIH research programs, Johns Hopkins University, and the FDA. Of interest to patients with ITP and platelet disorders was "FDA's Role in Rare Disease Research and Collaboration on the Path to Product Development" presented by Dr. Gayatri Rao, the Acting Director of the Office of Orphan Product Development at FDA. Dr. Rao said FDA is working to speed up approval of new drug treatments for rare diseases by use of its "Fast Track" and with priority review for serious or life-threatening diseases. In their Orphan Products Grants Program the FDA looks at animal models, clinical trials, and case studies to get the 'orphan product' designation. The research doesn't have to be random, double-blind tested initially, which helps speed the approval process for treatments.

The Director of the NIH National Heart Lung and Blood Institute (NHLBI), Dr. Jodi Black, discussed the NHLBI's Centers for Accelerated Innovations and VITA: Vascular Interventions/Innovations and Therapeutic Advances. The VITA program identifies, accelerates, and increases the number of highly innovative scientific discoveries that are translated into marketable treatment products. The VITA program addresses critical bottlenecks, decreases the time from discovery to product, increases the chances of success, encourages public-private partnerships, and fosters a culture for sustained technology development. For patients, this means new treatments for rare diseases like ITP can be found and approved sooner.

The afternoon session focused on other rare diseases, the NIH Undiagnosed Diseases Program, and presentations by patient group representatives from the National Organization of Rare Disorders (NORD) and the Genetic Alliance.

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