ICORD 2012- Abstract Form

36

THE MANTON CENTER FOR ORPHAN DISEASE RESEARCH: GENE DISCOVERY CORE

Objectives: The Manton Center for Orphan Disease Research is an interdisciplinary program established to provide the infrastructure needed to promote gene discovery in patients with orphan diseases, thereby leading to new diagnostic and therapeutic approaches for rare diseases. The Center's state-of-the-art "Gene Discovery Core" (GDC) aims to provide resources to investigators, including an IRB-approved protocol for patient/family enrollment, genetic studies, expertise, advice and logistical support and access to the latest research technologies, including Next Generation Sequencing (NGS).
Methods: The GDC links phenotypic data (i.e., medical and family history) to genomic data and biological samples (i.e. sequencing data, blood, tissues, etc.). Investigators can access samples and data for further analysis, and The Manton Center provides funding to support genetic studies on a competitive basis through internal grants. The GDC meets the need for a comprehensive protocol focused on enrolling and investigating patients with orphan diseases seen in clinic/in patient floors who are often lost to follow up.
Results: To date, investigators from more than 10 Divisions/Departments within Children's Hospital Boston have enrolled patients in the GDC, including the Divisions of Genetics, Gastroenterology and Newborn Medicine, and the Departments of Neurology, Urology and Cardiology. In total, 376 individuals, including those with rare or unknown disorders and their family members, have enrolled. Several individuals with strong evidence for a genetic disease, but without an identified genetic cause despite extensive clinical testing, have undergone NGS to identify the causative genetic alterations. To date, 96 individuals have been sent for analysis to identify the causative gene for the participant's health problems. In several cases, candidate genes have been identified and testing of family members, other affected individuals, and functional testing has begun. Importantly for the participants, the GDC has a mechanism to report research results back to the participants through The Manton Center genetic counselor and a health care provider of the participant's choosing.
Conclusions: The Manton Center GDC is a novel model that brings state-of-the-art research technologies to patients with rare diseases. The GDC reduces obstacles between the bedside and bench thereby helping to identify new disease conditions, etiologies and potential therapeutic targets. The GDC provides the infrastructure needed to efficiently carry out research on rare diseases.

- TITLES IN CAPITAL LETTERS, **Authors in bolds, Last name and Initials**, Institutions and e-mail in Arial normal size 11 letters. Provide text without free spaces, use international abbreviations or clarify them in the text and respect the space available in the form. The summary must clarify the objectives of the work, the used methods, the results and conclusions.
- The **deadline** for abstracts submission is **December 15th 2011.**