Online resource enables open data sharing for rare Mendelian diseases

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MyGene2, a new open data resource, helps patients with rare genetic conditions, clinicians, and researchers share information, connect with one another, and enable faster gene discovery, according to results presented at the American Society of Human Genetics (ASHG) 2017 Annual Meeting in Orlando, Fla.

"With MyGene2, we hope to help patients and their families engage with researchers and clinicians and vice versa, as well as speed up awareness of genes associated with rare Mendelian diseases," said Jessica Chong, PhD, Analysis Group Lead at the federally-funded University of Washington Center for Mendelian Genomics (UW-CMG), who presented the work.

Dr. Chong and Michael Bamshad, MD, Chief of the UW Division of Genetic Medicine, were working in rare disease gene discovery when they observed that a lack of open data sharing was slowing down scientific progress. "We found that good research findings were just not getting out there," she said. "Researchers may know of a gene's discovery years before it gets published, during which time patients and their families are undiagnosed and unaware."

At the same time, they noted that rare disease patients and families who were using existing social networks to make connections were running up against the limitations of those platforms, such as balancing worldwide reach with patient privacy and achieving consistency in terminology.

"We created MyGene2 to address these limitations, as a single place for patients and families to look for and share information rather than a hodgepodge of search terms and platforms," said Dr. Bamshad. The website helps patients enter their genetic data so it is searchable and useful to researchers and clinicians, along with detailed information about their phenotypes and experiences to help define symptoms and describe the course of disease. "This data can be de-identified to protect patient privacy, but remains complete and accessible, while allowing contact between families, researchers, and clinicians," Dr. Bamshad added.

In the year since the site was launched, more than 1,000 profiles have been created. The researchers are starting to see matches among patients with the same rare disease, and between patients and scientists studying their disease. In addition, they have been able to offer low-cost exome sequencing to patients whose insurance does not cover it, as well as comprehensive reporting of results and tools to identify secondary results.

Dr. Bamshad stresses the important role of patients and families in moving rare disease genetics forward. "Families need to be their own biggest advocates, and one way to do that is by sharing their data," he said.

More information: Chong JX et al. (2017 Oct 18). Abstract: Gene discovery via direct-to-family engagement using MyGene2. Presented at the American Society of Human Genetics 2017 Annual Meeting. Orlando, Florida. ep70.eventpilotadmin.com/web/p … =ASHG17&id=170121166

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