Interpreting clinical sequencing results for genome medicine

May 12 2016

Credit: CC0 Public Domain

This week, two important papers addressing pressing challenges and the best path forward for interpreting results from clinical genome and exome sequencing will appear in the *American Journal of Human*

Genetics.

One paper, "The Clinical Sequencing Exploratory Research (CSER) Consortium: Accelerating the Evidence-Based Practice of Genomic Medicine," highlights the accomplishments to date of more than 400 investigators who are working together to document their approaches for using sequencing to diagnose rare diseases in children and adults, diagnose and treat cancer, counsel couples who are planning to have children and more.

Dr. Robert C. Green, a medical geneticist and the lead author of the paper, is available to speak with members of the media to offer his perspective on the new Consortium and its goals.

A second paper also appearing in the *American Journal of Human Genetics*, titled "Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium," evaluates how shared rules and guidelines can help different labs interpret sequencing results consistently and resolve differences in interpretation.

Dr. Heidi Rehm, a clinical molecular geneticist and co-corresponding author of the paper, is available to share her views on variant interpretation guidelines and what they may mean for the future of clinical sequencing.

More information: *American Journal of Human Genetics*, DOI: 10.1016/j.ajhg.2016.04.011

Provided by Brigham and Women's Hospital