Non-Profit Patient Research Services from Rare Genomics Institute Enable Insight into Rare Diseases through Genome Sequencing and Cloud Collaboration

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Abstract

The non-profit Rare Genomics Institute (RG) helps provide rare disease patients with access to cutting-edge molecular biotechnology and analytical expertise with the potential to uncover new directions for research, treatment, and support. As volunteer analysts for RG, we analyzed the exome sequence data of patients with as-yet undiagnosed diseases with a suspected underlying genetic cause. Data analysis is performed with Omicia Opal, a web-based genome interpretation and reporting software platform integrating the machine learning algorithms VAAST and Phevor. We summarize our use of the Omicia Opal platform in three cases that represent the range of outcomes that can result from exome analysis; in one case we identified a likely pathogenic variant in the RDH12, associated with Leber's congenital amourosis; in a second case we identified a possibly pathogenic variant in IFT140, associated with Jeune syndrome; and in a third case we identified variants of uncertain significance in genes associated with Sotos and Weaver syndrome.

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