"The UAB Undiagnosed Diseases Program: Bioinformatic Applications in Rare Disorders"

Abstract:
The UAB Undiagnosed Diseases Program provides comprehensive diagnostic evaluations for children and adults with chronic, undiagnosed Disorders. Patients receive multidisciplinary evaluations, and most have whole genome sequencing (WGS) done as part of their evaluation. WGS is performed at HudsonAlpha Institute for Biotechnology along with bioinformatics evaluation. HudsonAlpha analysts meet monthly with clinicians at UAB to review results and decide which results to return to patients. Although sequencing is done on a research basis, any results that are returned are verified in a clinical laboratory, and a formal report is issued. In some cases, further functional genomic characterization is done, and data are shared through Genematcher or other components of Matchmaker Exchange. We are now working to develop an approach to therapeutic development for some instances where pathogenic variants are found that may be amenable to drug therapy.

Friday, 12/14/2018 at 10:15am
Shelby Biomedical Research Building, Room 105
1825 University Boulevard, Birmingham, AL 35294

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