Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

The Centers for Mendelian Genomics (CMGs, http://www.mendelian.org) aim to make major contributions to the discovery of the genetic basis of most or all Mendelian diseases in two main ways. The first is to use genome-wide sequencing (mostly whole exome sequencing) to discover the genetic basis of a large number of Mendelian traits during the funded period. The second is to accelerate the discovery by disseminating the obtained knowledge and effective approaches, reaching out to individual investigators, and coordinating with other rare disease programs worldwide.

Funding: 47 million (83% NHGRI and 17% NHLBI); Dec. 2011 – Dec. 2015

Funded centers

- Yale Center for Mendelian Diseases
- Baylor-Johns Hopkins Center for Mendelian Genetics
- University of Washington Center for Mendelian Genomics; Coordinating Center of Centers for Mendelian Genomics at University of Washington

Sample solicitation and public disease list: The CMGs solicit samples independently as well as jointly. The list of diseases under investigation by the CMGs is posted at http://www.mendelian.org/ and updated quarterly.

Outreach and international coordination

- The CMGs participate in the International Rare Diseases Research Consortium (IRDiRC, www.irdirc.org) in an effort to widely promote and coordinate rare disease gene discoveries. The Consortium members in Europe, Canada, and China are carrying out similar discovery efforts, currently at smaller scales.
- GeneMatcher (https://genematcher.org) has been made available to help solve unsolved cases by matching those cases. The site allows investigators to post genes of interest and will connect investigators who post the same gene.
- Since the inception of the program, the CMGs have collaborated with over 410 groups at 200 institutions in 33 countries.

Data release

- The causal variants published by the CMG can be browsed at http://data.mendelian.org/CMG/. These variants are displayed in custom tracks on UCSC Genome Browser.
- All CMGs deposit to dbGaP the data generated from samples appropriately consented for broad research use.

Discovery progress

- Over 80 publications have been made by the CMGs to date.
- As of January 31, 2014, the CMGs have sequenced over 10,000 exomes.