Mayo Clinic

Arizona — Scottsdale/Phoenix
Florida — Jacksonville
Minnesota — Rochester

Brief description of the "Clinical Center" or "Services on undiagnosed rare diseases"

Mayo Clinic is located in Rochester, MN, Jacksonville, FL and Scottsdale, AZ. Center for Individualized Medicine (CIM) at Mayo clinic has large number of physicians and scientists who work collaboratively on establishing diagnoses. The bioinformatics team is a critical part of our program. Program for undiagnosed rare disease was formally established in 2012, as a collaboration between CIM and Department of Clinical Genomics. The program evaluates children and adults by multidisciplinary clinical evaluation, laboratory testing, imaging and genomic evaluation. We have evaluated more than 1,000 cases so far, with 25% success in reaching diagnosis. Only in the last year we evaluated 500 patients. Several new variants in known disease causing genes have been identified and functionally confirmed as pathogenic. We linked previously unknown genes with specific phenotypes. Many discoveries were done in collaboration with other institutions.

Past (six months) and future/upcoming events related to undiagnosed rare disease. Please indicate date and venue.

Weekly case conference to review all cases and weekly Genome Odyssey Board meetings to discuss findings from genomic testing. Individualizing Medicine Conference (September 2017). Individualizing Medicine Conference (September 2018).

Relevant scientific publications (selected and / or published by the center)

1. Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. JIMD Rep. 2017 Oct 14. doi: 10.1007/8904_2017_59.
2. Novel NR2F1 variants likely disrupt DNA binding: molecular modeling in two cases, review of published cases, genotype-phenotype correlation, and phenotypic expansion of the Bosch-Boonstra-Schaaf optic atrophy syndrome. Cold Spring Harb Mol Case Stud. 2017 Nov 21;3(6) 3.
3. The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. Mol Genet Metab Rep. 2017, 11;13:46-51.
4. 4. A novel de novo frameshift deletion in EHMT1 in a patient with Kleefstra Syndrome results in decreased H3K9 dimethylation. Mol Genet Genomic Med. 2017, 26;5(2):141-146.

Grants. a) grant award to announce funding for projects; b) Received grants from other organizations/institutions for undiagnosed rare diseases research.

Center for Individualized Medicine funds several projects annually. Our team submitted the application to join UDN (NIH), pending decision.

Contact person

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