Undiagnosed Diseases Network

The Undiagnosed Diseases Network (UDN) is a research study funded by the National Institutes of Health Common Fund. The objectives of the UDN are to:

1. Improve the level of diagnosis and care for patients with undiagnosed diseases
2. Facilitate research into the etiology of undiagnosed diseases
3. Create an integrated and collaborative research community to identify improved options for optimal patient management

To learn more about the work of the UDN, check out the presentations below at ASHG 2017!

PLATFORM PRESENTATIONS

Session #6 // Wednesday // 10:15AM-10:30AM
10: Identification of an autosomal recessive form of Noonan Syndrome

Session #96 // Saturday // 9:00AM-9:15AM
331: Investigating non-coding variants: Characterizing potential disease-causing variants on microRNA binding sites

PLENARY PRESENTATION

Session #87 // Friday // 5:50PM-6:10PM
294: The Model Organisms Screening Center for the Undiagnosed Diseases Network

POSTER PRESENTATIONS

Wednesday // 2:00PM-3:00PM
523/W: Providing genomic medicine to the Hispanic population at the Stanford Center for Undiagnosed Diseases

Wednesday // 3:00PM-4:00PM
964/W: Characterizing the rare X-linked dominant variant in *ALG13*: A case report
1378/W: Quantitative assessment of the feasibility of using whole-genome sequencing data at common single nucleotide polymorphism positions to reproduce high-confidence genotype calling and copy number variation detection results from SNP microarray data

Thursday // 2:00PM-3:00PM
995/T: Heterozygous *COG4* variant causes a non-lethal type of COG4-CDG (formerly CDG-IIj)

Thursday // 3:00PM-4:00PM
1136/T: A growing need for reverse clinical genomics: Demonstrated by phenotypic characterization of *CDK13*-related disorders
932/T: Synonymous variant in *KCTD7* causes alternate splicing in siblings with progressive epilepsy

Friday // 11:30AM-12:30PM
1047/F: Mutations in *DDX3X* are a common cause of syndromic intellectual disability
951/F: 17p13.3 microdeletions between *YWHAE* and *LIS1* (*PAFAH1B1*) cause a unique leukoencephalopathy
2559/F: Covering all the bases: Case vignettes and diagnostic pipelines at the Stanford Center for Undiagnosed Diseases
2601/F: Undiagnosed Diseases Network (UDN) successes in precision medicine

Friday // 12:30PM-1:30PM
576/F: Effectiveness of a dedicated rotation in genomics, genetic medicine, and undiagnosed diseases for internal medicine residents
1416/F: Finding possible missed variant candidates through coverage comparison among varying sequencing technologies

ASHG 2017
Orlando, FL

NIH Common Fund Booth #952
NHGRI Booth #855

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