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!

**PLATF ORM 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-CGD (formerly CDG-Ii)

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

www.udnconnect.org
udn@hms.harvard.edu
1-844-746-4836 (RINGUDN)
udnconnect