Undiagnosed Diseases Network

ACMG 2018
Booth #720

The 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

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Thursday // 10:00AM-11:30AM
Poster Presentations - Odd Numbers

Abstract #231* *Vandana Shashi*
Resolving exome negative cases using an iterative approach results in high diagnosis rates

Abstract #239 *Tito Onyekweli*
GPI Anchor Disorders Demonstrate Abnormal Cholesterol Transport

Abstract #251 *Loren Pena*
Nonsense Variants in the Gene *IRF2BPL* are Associated with a Neurodegenerative Course

Abstract #317 *Heidi Cope*
Expansion of the Phenotype Associated with *EFL1*-Related Shwachman-Diamond Syndrome: Identification of a Patient with Short Stature, Metaphyseal Abnormalities and Thrombocytopenia

Abstract #343 *Devon Bonner*
Characterizing a de novo 5.75kb deletion in *ARID1B* missed by traditional genomic testing methods: a case report

Abstract #523* *Kelly Schoch*
My Patient Doesn't Have That! When Laboratory Results and Clinical Presentation are Discordant

Abstract #599* *Chris Lau*
Clinical Exome Sequence Analysis With Negative Outcome: Variant Re-assessment Strategies At the Undiagnosed Diseases Program

Abstract #671 *Alexander Moss*
An Examination of Undiagnosed Diseases Network Patient Demographics

Abstract #677 *Nikkola Carmichael*
Successfully Analyzed Rare Disease Cases From Brigham Genomic Medicine: What Case Characteristics Predispose to Resolution?

Abstract #683 *Cecilia Esteves*
Leveraging Online Social Networks to Increase Engagement in Rare Disease Research

Abstract #687 *John Phillips III*
Noncoding and Copy Number Variants Solve Multiple Undiagnosed Diseases Network (UDN) Mysteries

Abstract #747 *Thomas Markello*
Improving Completeness in Automated Agnostic Genome Wide Analysis, Ethnic Specific Priors and Automated Deleted Exon Detection

Friday // 10:30AM-12:00PM
Poster Presentations - Even Numbers

Abstract #226* *Donna Brown*
Research Reanalysis of Unsolved WGS Clinical Cases from the Undiagnosed Diseases Network

Abstract #234* *Jennefer Kohler*
A Multi-omics Approach to Interpretation of Copy Number Variants Identified Using Next-Generation Sequencing Data

Abstract #268 *Jill Rosenfeld*
Compound Heterozygous *TRIP11* Variants Cause a Non-lethal Form of Achondrogenesis Type 1A

Abstract #362 *Lauren Briere*
Phenotypic Variability in Early Infantile Epileptic Encephalopathy-44 caused by *UBA5* Mutations

Abstract #398 *Matt Holt*
Programmatic Detection of Diploid-Triploid Mixoploidy from Whole Genome Sequencing

Abstract #422 *Liliana Fernandez*
A New Case of Autosomal Dominant Fanconi Anemia, Complementation Group R in Association with a Novel, de novo *RAD51* Variant

Abstract #566 *Laurel Donnell-Fink*
Creating a Sustainable Clinical and Research Model for the Diagnosis of Rare Diseases

Abstract #654 *Kimberly LeBlanc*
Implementing a Patient Research Navigator (PRN) Process in the Undiagnosed Diseases Network (UDN)

Abstract #668 *Charlie Curnin*
LexiNV: A Pipeline for Analysis of Copy-Number Variants

Friday // 5:15PM-5:30PM
Platform Presentations - Genetic Counseling

Abstract #22 *Allyn McConkie-Rosell*
Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?

** Top rated poster presentation*

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