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.

**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* Jennifer 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