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.

**PLATFORM PRESENTATIONS**

**Wednesday 4:15 PM // Platform Presentations - Clinical Genetics and Therapeutics // 6E**
*8 Liz Worthey, HudsonAlpha Institute for Biotechnology*
Findings of the Whole Genome Sequencing Core of the Undiagnosed Diseases Network

**Thursday 8:45 AM // Featured Platform Presentations // 4E**
*28 Johannes Birngmeier, Stanford University*
ClinPhen Extracts and Prioritizes Patient Phenotypes Directly from Medical Records to Expedite Genetic Disease Diagnosis

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**ODD NUMBERED POSTERS**

**Thursday 10:00 AM – 11:30 AM**

**135 Camille Birch, HudsonAlpha Institute for Biotechnology**
Whole Genome Sequencing and Analysis of ME/CFS

**251* Kendall Burdick, Vanderbilt University Medical Center**
Limitations of Whole Exome Sequencing in Detecting Rare and Undiagnosed Diseases

**309 Joel Krier, Brigham and Women's Hospital**
Workflow, Implementation and Remaining Challenges for Reanalysis of Genomic Sequencing Data by a Clinical Genomics Program

**337 Diane Zastrow, Stanford University**
Compound Heterozygous Variants in IL6ST Associated with Immunodeficiency and GP130 Deficiency

**387 Tito Onyekwelu, NHGRI**
Oculodentodigital Dysplasia-associated GJA1 Mutation Leads to Deficiencies in CX43 Expression

**389 Jennefer Kohler, Stanford University**
Biallelic Variants in MRE11 Cause Ataxia-Telangiectasia-Like Disorder: A Case Report

**399 Laura Meissner, NHGRI**
Novel Variant Identified in DYRK1A-Related Intellectual Disability Syndrome by the Undiagnosed Diseases Program

**469 Liliana Fernandez, Stanford University**
A Novel, Pathogenic Variant in KMT2C in a Patient with Learning Disability, Cleft Palate, and Skeletal Abnormalities: A Case Report

**473 Donna Novacic, NHGRI**
Undiagnosed Diseases Network Clinical Case Report: Compound Heterozygous TOP3A Changes Manifest as a Mitochondrial Disease

**495 Devon Bonner, Stanford University**
DNASE1L3-related autoimmune disease: Case report and Molecular Profile

**523 Jeremy Woods, UCLA**
Myofibrillar Myopathy Associated with Homozygous PYROXD1

**603 Sho Yano, NHGRI**
Late-Onset Familial Episodic Aphasia with an Autosomal Dominant Inheritance Pattern

**693 Thomas Markello, NHGRI**
Automated Agnostic Genome Analysis Demonstrates a Net Difference Between Final Deleterious Candidate Lists of Probands Versus Unaffected Siblings Analyzed Symmetrically

**739 Christopher Lau, NHGRI**
Reanalysis of Negative Clinical Exome in Undiagnosed Diseases: Assessing the Level of Evidence and Clinical Validity of Gene-Disease Associations

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**EVEN NUMBERED POSTERS**

**FRIDAY 10:30 AM – 12:00 PM**

**116 Harish Chatrathi, NHGRI**
Novel De Novo CUL3 Mutation in a Patient with Gordon’s Syndrome Results in Altered Function of Cullin-RING E3 Ubiquitin Ligase

**328 Jeremy Woods, UCLA**
Microtubule abnormalities and mitochondrial network dysfunction in mitochondrial myopathy and ataxia associated with pathogenic variants in MSTO1

**346 Jill Rosenfeld (Mokry), Baylor College of Medicine**
Overcoming the “N of 1” Problem: Novel Disease Gene Discovery in the Undiagnosed Diseases Network

**352 Nadiya Sosonkina, HudsonAlpha Institute for Biotechnology**
A Finding in Whole Genome Sequencing of an Individual with Undiagnosed Disease Suggests an Ethnicity-Specific Gene Duplication Event

**452 Elly Brokamp, Vanderbilt University Medical Center**
Evidence for a New MSL2-Related Disease Using Internal VUMC De-Identified Database

**508 Colleen Evans, NHGRI**
Recurrent de novo SPG4 Mutation Causes an Atypical Phenotype of Severe Progressive Early-onset Spastic Quadriaparesis in Two Unrelated Individuals

**536 Linnea Westerkam, NHGRI**
The Importance of Exploring Multiple Genetic Explanations as Demonstrated by a Blended Phenotype of EHMT1 and ACAN Variants

**580 Marta Maria Majcherska, Stanford University**
Unusual Cardiac Presentations at the Stanford Center for Undiagnosed Diseases

**744 Kyle Reichard, NHGRI**
The characterization of a novel zebrafish model for a human seizure disorder caused by mutations in PRUNE1

**800 Hongzheng Dai, Baylor Genetics**
A common pan-ethnic exonic deletion in TBCK gene causes early onset hypotonia and psychomotor retardation identified through clinical exome sequencing

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