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 Birnmeier, Stanford University
ClinPhen Extracts and Prioritizes Patient Phenotypes Directly from Medical Records to Expedite Genetic Disease Diagnosis

**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 ILEST Associated with Immunodeficiency and GP130 Deficiency

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

389 Jennifer 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

**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 MSTR1

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 Linneua 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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