

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 Birgmeier, 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 UniversityCompound Heterozygous Variants in *IL6ST* Associated with Immunodeficiency and GP130 Deficiency**387 Tito Onyekweli, 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* Pathogenic Variants Detected by Exome Sequencing**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 Proband 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 *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 Ely Brokamp, Vanderbilt University Medical CenterEvidence 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 Quadriplegia 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, NHGRIThe 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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