Undiagnosed Diseases Network

ACMG 2019
Booth #201

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

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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 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 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 *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 Quadriparesis 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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