Undiagnosed Diseases Network

ASHG 2018
NHGRI Booth #329

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.

TALKS

Wed Oct 17, 9:30AM // Concurrent Platform Session A, 12. Bone and Muscle: Identifying Causal Genes
31 *Ronit Marom, Baylor College of Medicine*
COPB2 loss of function leads to disrupted collagen trafficking and juvenile osteoporosis

Wed Oct 17, 6:00PM // Concurrent Platform Session C, 34. Reanalysis of Sequencing Data to Increase Diagnostic Yield
99 *Christopher Lau, NHGRI*
Assessing variants in genes of unknown significance: the quest for novel gene discoveries at the NIH Undiagnosed Diseases Program

Fri Oct 19, 9:00AM // Concurrent Platform Session F, 69. Using RNA-seq to Improve DNA Sequence Interpretation
255 *Shan Chen, Baylor College of Medicine*
Rare disease diagnosis by integrating RNA sequencing in the Undiagnosed Diseases Network

Fri Oct 19, 5:40PM // Featured Plenary Abstract Session II
277 *Lindsay Burrage, Baylor College of Medicine*
Biallelic variants in *TONSL* cause SPONASTRIME dysplasia and an expanded spectrum of skeletal dysplasia phenotype

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POSTERS

Wed Oct 17, 2:00-3:00PM

1383 *Hane Lee, UCLA*
Identifying splice alterations using RNAseq to improve genetic diagnosis for rare Mendelian disorders

1797 *Diane Zastrow, Stanford University*
Beyond the exome report: approaches to additional analysis for undiagnosed genetic disease

3003* *Xia Wang, Baylor College of Medicine*
De novo missense variants in *TRAF7* cause developmental delay, congenital anomalies, and dysmorphic features

3093* *John Phillips III, Vanderbilt University*
Familial autonomic ganglionopathy and neurogenic orthostatic hypotension associated with rare *CHRNA3* variants

Wed Oct 17, 3:00-4:00PM

1194 *Devon Bonner, Stanford University*
Dilated cardiomyopathy: a novel finding in a patient with *ADSSL1*-related myopathy

Thu Oct 18, 2:00-3:00PM

1207 *Jennefer Kohler, Stanford University*
Non-coding variants in *MECR*: case report and molecular phenotype

3055 *David Murdock, Baylor College of Medicine*
Comparative quality of whole exome sequencing among commercial laboratories for patients in the Undiagnosed Diseases Network

Fri Oct 19, 2:00-3:00PM

1535 *Nick Balanda, NHGRI*
Transcriptome analysis by RNA-sequencing as an adjunct to whole genome analysis in undiagnosed genetic disease

Fri Oct 19, 3:00-4:00PM

1112 *Liliana Fernandez, Stanford University*
A new case of an intermediate phenotype along the spectrum of *ATP1A3*-related neurological disorders

1412 *Blythe Hospelhorn, NHGRI*
Construction of a structural variant detection pipeline for the Undiagnosed Diseases Program

1580* *Shruti Marwaha, Stanford University*
Comparison of indel callers and metrics to evaluate performance of new tools

1682 *Daron Ross, NHGRI*
Undiagnosed Diseases Program Database (UDPdb) of genomic data: development and utility

2894 *Jeremy Woods, UCLA*
Characterization of recessive myopathy and ataxia syndrome due to *MSTO1* variants

*** Reviewer's Choice Abstract***
