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**


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 TONSOL cause SPONASTRIME dysplasia and an expanded spectrum of skeletal dysplasia phenotype

**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