Undiagnosed Diseases Network (UDN) talks and posters at the ASHG 2020 Virtual Meeting

PLATFORM PRESENTATIONS

SESSION 005 - Molecular and Cytogenetic Diagnostics: What have we been missing?
Tuesday // October 27, 2020 // 12:00 PM – 12:15 PM 1032 - Transcriptome-directed genomic analysis accelerates diagnosis and overcomes limitations of exome sequencing and chromosomal microarray (D.R. Murdock)

SESSION 008 - Featured Plenary Abstract Session I
Tuesday // October 27, 2020 // 1:00 PM - 1:20 PM 1052 - Somatic Mutations in a Single Residue of *UBA1* are Associated with a Severe Adult-Onset Autoinflammatory Disease (D. Beck)

SESSION 013 - Machine Learning
Wednesday // October 28, 2020 // 11:15 AM - 11:30 AM 1118 - Identifying rare variant combinations associated with digenic disease in Undiagnosed Diseases Network patients using machine learning (S. Mukherjee)

SESSION 039 - Computational approaches for disease diagnosis and variant effect determination
Friday // October 30, 2020 // 11:00 AM - 11:15 AM 1300 - Personalized regulatory genomics: Identifying gene dysregulation to solve undiagnosed rare disease cases (E. McArthur)

POSTER TALK

SESSION 036 - Poster Talks
Tuesday // October 27, 2020 // 5:39 PM - 5:42 PM 1088 - Bi-allelic loss-of-function variants in Replication Factor C 4 (*RFC4*) are associated with a neurological disorder characterized by ataxia and incoordination (M. Morimoto)

ON-DEMAND POSTERS

SESSION 209 - Molecular and Cytogenetic Diagnostics
3306 - Clinical whole genome sequencing increases molecular diagnostic yields of Mendelian disorders in patients with prior negative exome evaluations (P. Liu)
3325 - Establishing molecular diagnoses for rare monogenic disease: Why do we fail? (J. Krier)
3357 - Neurodevelopmental disorders with myoclonic epilepsy in the NIH Undiagnosed Diseases Program (S. Shimada)

SESSION 208 - Mendelian Phenotypes
3089 - 8 year-old girl with hypokalemic periodic paralysis and SOFT syndrome: Novel, disease-associated variants identified in *SCN4A* and *POC1A* (L. Fernandez)
3137 - Clinical sites of the Undiagnosed Diseases Network provide unique contributions to genomic medicine and science. (V. Shashi)
3157 - Dysregulation of protein synthesis due to loss of function of *PUS7* is associated with postnatal microcephaly, aggressive self-injurious behavior, speech delay, and global developmental delay (S. Han)
3191 - *HIP1R* may be associated with a recessive, progressive, neurologic disorder (J. A. Rosenfeld)
3201 - Infantile-onset obesity, tall stature, and macrocephaly: diagnosing atypical presentations of STX16 microdeletion-associated pseudohypoparathyroidism type 1B by genome sequencing in two sisters (A. Xu)
3206 - Long read genome sequencing identifies a novel inherited multi-exon duplication of *EHMT1* in a patient with likely Kleefstra syndrome: A case of parental mosaicism? (C. Reuter)
3234 - Predicted loss-of-function variants in *SNAPC4* are associated with a pediatric neurological disorder characterized by progressive neuromuscular spasticity, developmental delay, and speech dysarthria (F. G. Frost)

SESSION 211 - Molecular Phenotyping and Omics Technologies
3491 - Assessing the utility of RNA sequencing in the diagnosis of rare Mendelian diseases. (D. E. Bonner)

SESSION 201 - Bioinformatics and Computational Approaches
2027 - Algorithm with O(N) Time Complexity for Detecting Mosaicism in DNA Samples with SNP Chip and Exome Sequencing (A. Rodriguez)
2148 - Leveraging whole genome sequencing (WGS) data in search of known and novel repeat expansion diseases in the Undiagnosed Diseases Program (UDP) (P. Houston)
2168 - NIH Undiagnosed Diseases Program data sharing, part II. Tracking case matches (D. Adams)
2179 - Platform Independent Distributed Model for the Visualization of Exome and Genome Analysis Results (B. N. Pusey)

COVID-19 update: The UDN is closely monitoring the ongoing coronavirus disease 2019 (COVID-19) pandemic. We are taking informed action to limit the spread of COVID-19 while ensuring the continuity of our research mission. **The UDN clinical sites remain fully staffed and continue to review new applications.** In-person evaluations may be delayed or replaced with virtual evaluations in accordance with hospital and university policies. For more information, please visit our website **udnconnect.org**.