Undiagnosed Diseases Network (UDN) talks and posters at the ASHG 2021 Virtual Meeting

PLATFORM PRESENTATIONS

Cutting-edge approaches and novel gene discovery in neurodevelopmental disorders | October 20 | 10:30 AM – 12:00 PM ET

1153 - Bi-allelic variants in the autophagy gene *ATG4D* are associated with a pediatric neurological disorder characterized by hypotonia, joint laxity, and delayed speech and motor development (M. Morimoto)

1156 - Statistical framework uncovers deep intronic splice gain variants implicated in undiagnosed cases (S. Kobren)

Multidisciplinary omics approaches for diagnostics | October 20 | 10:30 AM – 12:00 PM (Eastern)

1187 - Large scale Cas9 mediated depletion of highly abundant transcripts to expand the interpretable genome and improve the diagnostic yield of clinical RNA Seq (A. Y. Huang)

POSTER PRESENTATIONS

SESSION A | October 18 | 9:30 AM - 10:30 AM ET

2186 - Bi-allelic variants in neuronal cell adhesion molecule (*NRCAM*) lead to a novel neurodevelopmental disorder characterized by developmental delay, hypotonia, peripheral neuropathy or spasticity (H. Baris Feldman)

SESSION B | October 18 | 11:30 AM - 12:30 PM ET

2550 - De novo variants in *TCF4* with a suspected gain-of-function mechanism are responsible for a new malformative disease without intellectual disability (E. Colin)

2630 - Pathogenic *SPTBN1* variants cause an autosomal dominant neurodevelopmental syndrome (M. A. Cousin)

SESSION C | October 18 | 1:30 PM - 2:30 PM ET

2891 - Prioritizing Research Variants in the NIH Undiagnosed Diseases Program (D. R. Adams)

3002 - Characterizing repeat expansion variation in the Undiagnosed Disease Network cohort (S. Fazal)

3037 - Impaired *SNAPC4* function leads to global reduction of canonical splicing events and is associated with a disorder characterized by progressive spasticity, developmental delay, and speech dysarthria (F. G. Frost)

3104 - X-linked recessive mutations in *PDZD4* are likely associated with neurodevelopment delay and autism spectrum disorder (M. Sandler)

SESSION D | October 18 | 3:30 PM - 4:30 PM ET

3410 - De novo damaging variants in the microRNA processor *DROSHA* are associated with a severe progressive neurological disorder (S. Barish)

3411 - De novo variant in *MRTF-B* is associated with intellectual disability, minor dysmorphic features, expressive language delay, impulse control issues, and fine motor delay (J. C. Andrews)

3417 - Biallelic *NAV2* truncating variants cause a neurodevelopmental disorder with cerebellar cortical dysplasia (J. A. Rosenfeld)

3426 - Loss of function variants in *PUS7* dysregulate protein synthesis and are associated with neurodevelopmental delay, microcephaly, and hyperuricemia (K. Garcia)

3428 - Novel *PRUNE1* c.933G>A (p.Thr311=) synonymous splice variant induces exon 7 skipping and leads to an atypical presentation of NMIHBA syndrome: Case report and review of the literature (C. Magyar)

3435 - A heterozygous gain-of-function variant in *KIF5B* causes osteogenesis imperfecta by disrupting the Golgi-primary cilia axis (M. Washington)

3456 - Precision medicine models for undiagnosed and rare disease (L. C. Burrage)

3482 - Personalized structural biology reveals the molecular mechanisms underlying heterogeneous epileptic phenotypes caused by de novo *KCNC2* variants (S. Mukherjee)

3502 - Knowledge based artificial intelligence for variant pathogenicity prediction for Mendelian disorders (D. Mao)