Tarja Linnankivi
List of Publications 1/2014- 11/2018

Original articles

Heyne HO, Singh T, Stamberger H, et al. De novo variants in neurodevelopmental disorders with epilepsy. Nat Genet. 2018 Jul;50(7)

Johannesen KM, Gardella E, **Linnankivi T**, et al. Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia 2018;59:389-402.

Allen, A. S, Berkovic, S. F, Bridgers, J, et al. Application of rare variant transmission disequilibrum tests to epileptic encephalopathy trio sequence data. Eur J Hum Genet 2017;25, 7:894-899.

Platzer K, Yuan H, Schütz H, et al. *GRIN2B* encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. J Med Genet 2017; 54:460-470.

Muona M, Ishimura R, Laari A, Ichimura Y, **Linnankivi T**, et al. Biallelic Variants in UBA5 Link Dysfunctional UFM1 Ubiquitin-like Modifier Pathway to Severe Infantile-Onset Encephalopathy. Am J Hum Genet 2016;99:683-94

Linnankivi T, Neupane N, Richter U, et al. Splicing defect in mitochondrial seryl-tRNA synthetase gene cause progressive spastic paresis instead of HUPRA syndrome. Hum Mut 2016;37:884-8.

Crow YJ, Chase DS, Lowenstein Schmidt J, et al. Characterization of human disease phenotypes associated with mutation in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR and IFIH1. Am J Med Genet A. 2015;167A:292-312.

Syrbe S, Hedrich UB, Riesch E, et al. De novo loss- or gain-of-function mutation in KCNA2 cause epileptic encephalopathy. Nat Genet 2015;47:393-9.

Anttonen EK, Hilander T, **Linnankivi T**, et al. Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. Neurology 2015; 85:306-15.

Appenzeller S, Balling R, Barisic N, et al. De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. Am J Hum Genet 2014;95:360-70.

Van Berge L, Hamilton EM, **Linnankivi T**, et al. Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation; clinical and genetic characterization and target for therapy. Brain 2014; 137:1019-29.

Original articles and reviews in Finnish textbooks and refereed publication series

Linnankivi T. Neuroimmunologiaa. Kirjassa: Pihko H, Haataja L, Rantala H, (eds). Lastenneurologia. Helsinki: Duodecim, 2014.

Popularized articles

Muona M, **Linnankivi T**, Lehesjoki A-E. Geenit ja epilepsia. Epilepsialehti 2014; 45:6-8.