Sibel Aylin Ugur

List of Publications by Year
in descending order

14 papers	100 citations	1307594 7 h-index	1474206 9 g-index
14 all docs	14 docs citations	14 times ranked	175 citing authors

#	Article	IF	Citations
1	Two cases with mitochondrial membrane protein-associated neurodegeneration: genetic features and long-term clinical follow-up. Neurocase, 2022, , 1-5.	0.6	1
2	Hypomyelinating spastic dyskinesia and ichthyosis caused by a homozygous splice site mutation leading to exon skipping in ELOVL1. Brain and Development, 2022, 44, 391-400.	1.1	8
3	The rare rs769301934 variant in NHLRC1 is a common cause of Lafora disease in Turkey. Journal of Human Genetics, 2021, 66, 1145-1151.	2.3	2
4	Effect of the brain-derived neurotrophic factor gene Val66Met polymorphism on sensory-motor integration during a complex motor learning exercise. Brain Research, 2020, 1732, 146652.	2.2	17
5	A Novel and Mosaic WDR45 Nonsense Variant Causes Beta-Propeller Protein-Associated Neurodegeneration Identified Through Whole Exome Sequencing and X chromosome Heterozygosity Analysis. NeuroMolecular Medicine, 2019, 21, 54-59.	3.4	5
6	Identification of epilepsy related pathways using genome-wide DNA methylation measures: A trio-based approach. PLoS ONE, 2019, 14, e0211917.	2.5	6
7	Biallelic loss of EEF1D function links heat shock response pathway to autosomal recessive intellectual disability. Journal of Human Genetics, 2019, 64, 421-426.	2.3	13
8	Clinical and genetic spectrum of an orphan disease MPAN: a series with new variants and a novel phenotype. Neurologia I Neurochirurgia Polska, 2019, 53, 476-483.	1.2	7
9	Clinical phenotype of hereditary spastic paraplegia due to KIF1C gene mutations across life span. Brain and Development, 2018, 40, 458-464.	1.1	13
10	A novel homozygous GALC variant has been associated with Krabbe disease in a consanguineous family. Neurological Sciences, 2018, 39, 2123-2128.	1.9	0
11	SYNE1 related cerebellar ataxia presents with variable phenotypes in a consanguineous family from Turkey. Neurological Sciences, 2017, 38, 2203-2207.	1.9	8
12	A novel gene mutation in PANK2 in a patient with severe jaw-opening dystonia. Brain and Development, 2016, 38, 755-758.	1.1	7
13	Screening LGI1 in a cohort of 26 lateral temporal lobe epilepsy patients with auditory aura from Turkey detects a novel de novo mutation. Epilepsy Research, 2016, 120, 73-78.	1.6	5
14	A clinical variant in SCN1A inherited from a mosaic father cosegregates with a novel variant to cause Dravet syndrome in a consanguineous family. Epilepsy Research, 2015, 113, 5-10.	1.6	8