Gali Heimer

List of Publications by Year
in descending order

27	1,047	706676 14	591227 27
papers	citations	h-index	g-index

27	27	27	2863
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.5	11
2	A tecpr2 knockout mouse exhibits age-dependent neuroaxonal dystrophy associated with autophagosome accumulation. Autophagy, 2021, 17, 3082-3095.	4.3	18
3	Lysosomal targeting of autophagosomes by the TECPR domain of TECPR2. Autophagy, 2021, 17, 3096-3108.	4.3	20
4	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
5	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	0.7	13
6	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	1.1	18
7	Delineation of the phenotype of MED17-related disease in Caucasus-Jewish families. European Journal of Paediatric Neurology, 2021, 32, 40-45.	0.7	3
8	A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. Scientific Reports, 2021, 11, 19099.	1.6	13
9	Development of an adapted Clinical Global Impression scale for use in Angelman syndrome. Journal of Neurodevelopmental Disorders, 2021, 13, 3.	1.5	12
10	The STARS Phase 2 Study. Neurology, 2021, 96, e1024-e1035.	1.5	12
11	Personalized treatment with retigabine for pharmacoresistant epilepsy arising from a pathogenic variant in the KCNQ2 selectivity filter. Epileptic Disorders, 2021, 23, 695-705.	0.7	14
12	Netrinâ€G2 dysfunction causes a Rettâ€like phenotype with areflexia. Human Mutation, 2020, 41, 476-486.	1.1	10
13	Functional parameter measurements in children with ataxia telangiectasia. Developmental Medicine and Child Neurology, 2020, 62, 207-213.	1.1	4
14	Four patients with D-bifunctional protein (DBP) deficiency: Expanding the phenotypic spectrum of a highly variable disease. Molecular Genetics and Metabolism Reports, 2020, 25, 100631.	0.4	11
15	In the eye of the beholder: Using a multiple-informant approach to examine the mediating effect of cognitive functioning on emotional and behavioral problems in children with an active epilepsy. Seizure: the Journal of the British Epilepsy Association, 2020, 82, 31-38.	0.9	2
16	Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. American Journal of Human Genetics, 2020, 106, 246-255.	2.6	17
17	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
18	Cerebral and portal vein thrombosis, macrocephaly and atypical absence seizures in Glycosylphosphatidyl inositol deficiency due to a PIGM promoter mutation. Molecular Genetics and Metabolism, 2019, 128, 151-161.	0.5	9

#	Article	IF	Citations
19	Loss of Protocadherinâ€12<scp>L</scp>eads to <scp>D</scp>iencephalicâ€<scp>M</scp>esencephalic <scp>J</scp>unction <scp>D</scp>ysplasia <scp>S</scp>yndrome. Annals of Neurology, 2018, 84, 638-647.	2.8	19
20	Secondary enuresis and urological manifestations in children with ataxia telangiectasia. European Journal of Paediatric Neurology, 2018, 22, 1118-1123.	0.7	1
21	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127
22	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. Brain, 2017, 140, 568-581.	3.7	53
23	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	2.6	43
24	Influence of epileptic activity during sleep on cognitive performance in benign childhood epilepsy with centrotemporal spikes. European Journal of Paediatric Neurology, 2017, 21, 858-863.	0.7	19
25	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	2.6	91
26	TECPR2 mutations cause a new subtype of familial dysautonomia like hereditary sensory autonomic neuropathy with intellectual disability. European Journal of Paediatric Neurology, 2016, 20, 69-79.	0.7	45
27	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. Genetics in Medicine, 2015, 17, 774-781.	1.1	284