Natalie S Hauser

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1275166/publications.pdf

Version: 2024-02-01

22	583	758635 12	676716 22
papers	citations	h-index	g-index

23	23	23	1453
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American Journal of Human Genetics, 2016, 99, 711-719.	2.6	81
2	FOXG1 syndrome: genotypeâ€“phenotype association in 83 patients with FOXG1 variants. Genetics in Medicine, 2018, 20, 98-108.	1.1	77
3	De novo missense variants in PPP2R5D are associated with intellectual disability, macrocephaly, hypotonia, and autism. Neurogenetics, 2016, 17, 43-49.	0.7	61
4	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	1.8	59
5	Regions of homozygosity identified by oligonucleotide SNP arrays: evaluating the incidence and clinical utility. European Journal of Human Genetics, 2015, 23, 663-671.	1.4	54
6	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	2.8	42
7	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	0.7	34
8	Endosomal trafficking defects in patient cells with KIAA1109 biallelic variants. Genes and Diseases, 2019, 6, 56-67.	1.5	22
9	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64.	1.6	19
10	Pitfalls of clinical exome and gene panel testing: alternative transcripts. Genetics in Medicine, 2019, 21, 1240-1245.	1.1	17
11	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	1.1	14
12	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.0	14
13	Genomic analysis of an infant with intractable diarrhea and dilated cardiomyopathy. Journal of Physical Education and Sports Management, 2017, 3, a002055.	0.5	13
14	Experience with genomic sequencing in pediatric patients with congenital cardiac defects in a large community hospital. Molecular Genetics & Genomic Medicine, 2018, 6, 200-212.	0.6	12
15	Genotype-first analysis of a generally healthy population cohort supports genetic testing for diagnosis of hereditary angioedema of unknown cause. Allergy, Asthma and Clinical Immunology, 2019, 15, 32.	0.9	12
16	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. Brain, 2019, 142, e15-e15.	3.7	12
17	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	0.7	12
18	Mutation in an alternative transcript of CDKL5 in a boy with early-onset seizures. Journal of Physical Education and Sports Management, 2018, 4, a002360.	0.5	10

#	Article	IF	Citations
19	Mosaicism in ASXL3-related syndrome: Description of five patients from three families. European Journal of Medical Genetics, 2020, 63, 103925.	0.7	9
20	EPG5 Variants with Modest Functional Impact Result in an Ameliorated and Primarily Neurological Phenotype in a 3.5-Year-Old Patient with Vici Syndrome. Neuropediatrics, 2019, 50, 257-261.	0.3	5
21	A case study of atypical Larsen syndrome with absent hallmark joint dislocations. Molecular Genetics & Genomic Medicine, 2019, 7, e648.	0.6	2
22	Diagnosis of LCHAD/TFP deficiency in an at risk newborn using umbilical cord blood acylcarnitine analysis. Molecular Genetics and Metabolism Reports, 2017, 10, 8-10.	0.4	1