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	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
2	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
3	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â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in 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
4	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	1.1	14
5	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	2.8	42
6	Mosaicism in ASXL3-related syndrome: Description of five patients from three families. European Journal of Medical Genetics, 2020, 63, 103925.	0.7	9
7	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
8	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
9	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. Brain, 2019, 142, e15-e15.	3.7	12
10	Endosomal trafficking defects in patient cells with KIAA1109 biallelic variants. Genes and Diseases, 2019, 6, 56-67.	1.5	22
11	A case study of atypical Larsen syndrome with absent hallmark joint dislocations. Molecular Genetics & Genomic Medicine, 2019, 7, e648.	0.6	2
12	Pitfalls of clinical exome and gene panel testing: alternative transcripts. Genetics in Medicine, 2019, 21, 1240-1245.	1.1	17
13	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64.	1.6	19
14	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
15	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
16	FOXG1 syndrome: genotypeâ€“phenotype association in 83 patients with FOXG1 variants. Genetics in Medicine, 2018, 20, 98-108.	1.1	77
17	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
18	Genomic analysis of an infant with intractable diarrhea and dilated cardiomyopathy. Journal of Physical Education and Sports Management, 2017, 3, a002055.	0.5	13

#	Article	IF	Citations
19	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
20	De novo missense variants in PPP2R5D are associated with intellectual disability, macrocephaly, hypotonia, and autism. Neurogenetics, 2016, 17, 43-49.	0.7	61
21	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	1.8	59
22	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