

Julie S Cohen

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

1,407
citations

394421

19
h-index

361022

35
g-index

40
all docs

40
docs citations

40
times ranked

3430
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical whole exome sequencing in child neurology practice. <i>Annals of Neurology</i> , 2014, 76, 473-483.	5.3	228
2	Mutations disrupting neurogenesis genes confer risk for cerebral palsy. <i>Nature Genetics</i> , 2020, 52, 1046-1056.	21.4	96
3	MEF2C Haploinsufficiency features consistent hyperkinesia, variable epilepsy, and has a role in dorsal and ventral neuronal developmental pathways. <i>Neurogenetics</i> , 2013, 14, 99-111.	1.4	89
4	A Diagnostic Approach for Cerebral Palsy in the Genomic Era. <i>NeuroMolecular Medicine</i> , 2014, 16, 821-844.	3.4	89
5	FOXP1 syndrome: genotype-phenotype association in 83 patients with FOXP1 variants. <i>Genetics in Medicine</i> , 2018, 20, 98-108.	2.4	77
6	RANBP2 and CLTC are involved in ALK rearrangements in inflammatory myofibroblastic tumors. <i>Cancer Genetics and Cytogenetics</i> , 2007, 176, 107-114.	1.0	72
7	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. <i>Journal of Clinical Investigation</i> , 2019, 129, 1240-1256.	8.2	68
8	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. <i>American Journal of Human Genetics</i> , 2020, 107, 352-363.	6.2	64
9	<i>ELP2</i> is a novel gene implicated in neurodevelopmental disabilities. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1391-1395.	1.2	61
10	Development and validation of the Psychological Adaptation Scale (PAS): Use in six studies of adaptation to a health condition or risk. <i>Patient Education and Counseling</i> , 2013, 93, 248-254.	2.2	54
11	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. <i>American Journal of Human Genetics</i> , 2017, 101, 1013-1020.	6.2	53
12	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2019, 105, 413-424.	6.2	43
13	SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. <i>Human Mutation</i> , 2019, 40, 908-925.	2.5	39
14	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. <i>European Journal of Human Genetics</i> , 2016, 24, 1761-1770.	2.8	36
15	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. <i>PLoS Genetics</i> , 2017, 13, e1006683.	3.5	35
16	Clinical whole-exome sequencing results impact medical management. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 1068-1078.	1.2	33
17	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	6.2	30
18	ST3GAL5-Related Disorders: A Deficiency in Ganglioside Metabolism and a Genetic Cause of Intellectual Disability and Choreoathetosis. <i>Journal of Child Neurology</i> , 2018, 33, 825-831.	1.4	28

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19	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. <i>Nature Communications</i> , 2021, 12, 2678.	12.8	26
20	Constitutional Duplication of a Region of Chromosome Yp Encoding AMELY, PRKY, and TBL1Y. <i>Journal of Molecular Diagnostics</i> , 2007, 9, 408-413.	2.8	23
21	Genetic Counseling in Neurodevelopmental Disorders. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2020, 10, a036533.	6.2	20
22	Posterior Neocortex-Specific Regulation of Neuronal Migration by CEP85L Identifies Maternal Centriole-Dependent Activation of CDK5. <i>Neuron</i> , 2020, 106, 246-255.e6.	8.1	19
23	Compound Heterozygous Variants in ROBO1 Cause a Neurodevelopmental Disorder With Absence of Transverse Pontine Fibers and Thinning of the Anterior Commissure and Corpus Callosum. <i>Pediatric Neurology</i> , 2017, 70, 70-74.	2.1	16
24	The variability of <i>SMARCA4</i> -related Coffin-Siris syndrome: Do nonsense candidate variants add to milder phenotypes?. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2058-2067.	1.2	14
25	Expansion of the genetic landscape of <i>ERLIN2</i> -related disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 573-578.	3.7	12
26	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. <i>American Journal of Human Genetics</i> , 2021, 108, 2006-2016.	6.2	11
27	Challenges to informed consent for exome sequencing: A best-worst scaling experiment. <i>Journal of Genetic Counseling</i> , 2019, 28, 1189-1197.	1.6	7
28	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. <i>Brain Sciences</i> , 2021, 11, 931.	2.3	7
29	The clinical and molecular spectrum of <i>QRICH1</i> associated neurodevelopmental disorder. <i>Human Mutation</i> , 2022, 43, 266-282.	2.5	7
30	Mitochondrial DNA Deletions With Low-Level Heteroplasmy in Adult-Onset Myopathy. <i>Journal of Clinical Neuromuscular Disease</i> , 2018, 19, 117-123.	0.7	6
31	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. <i>Brain</i> , 2020, 143, e31-e31.	7.6	6
32	Successful treatment of choreo-athetotic movements in a patient with an EEF1A2 gene variant. <i>SAGE Open Medical Case Reports</i> , 2018, 6, 2050313X1880762.	0.3	5
33	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. <i>Genetics in Medicine</i> , 2021, 23, 1873-1881.	2.4	5
34	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2021, 23, 1912-1921.	2.4	5
35	Novel variants in <i>KAT6B</i> spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1809.	1.2	4
36	Diagnostic Evaluation in Children With Developmental Delay. <i>Clinical Pediatrics</i> , 2012, 51, 1208-1210.	0.8	3

#	ARTICLE	IF	CITATIONS
37	Adult patients with undiagnosed conditions and their responses to unresolved uncertainty from exome sequencing. <i>Journal of Genetic Counseling</i> , 2020, 29, 992-1003.	1.6	3
38	Vigabatrin as a Targeted Treatment of GABAB Receptor-Related Epileptic Encephalopathy. <i>Pediatric Neurology</i> , 2019, 99, 82-84.	2.1	2