Julie S Cohen

List of Publications by Year
in descending order

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	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
2	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. Nature Communications, 2021, 12, 2678.	12.8	26
3	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	2.4	5
4	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	2.4	5
5	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. Brain Sciences, 2021, 11, 931.	2.3	7
6	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	1.2	4
7	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
8	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	21.4	96
9	The variability of <scp>SMARCA4</scp>â€related <scp>Coffinâ€“Siris</scp> syndrome: Do nonsense candidate variants add to milder phenotypes?. American Journal of Medical Genetics, Part A, 2020, 182, 2058-2067.	1.2	14
10	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. American Journal of Human Genetics, 2020, 107, 352-363.	6.2	64
11	Expansion of the genetic landscape of ERLIN2â€related disorders. Annals of Clinical and Translational Neurology, 2020, 7, 573-578.	3.7	12
12	Adult patients with undiagnosed conditions and their responses to unresolved uncertainty from exome sequencing. Journal of Genetic Counseling, 2020, 29, 992-1003.	1.6	3
13	Posterior Neocortex-Specific Regulation of Neuronal Migration by CEP85L Identifies Maternal Centriole-Dependent Activation of CDK5. Neuron, 2020, 106, 246-255.e6.	8.1	19
14	Genetic Counseling in Neurodevelopmental Disorders. Cold Spring Harbor Perspectives in Medicine, 2020, 10, a036533.	6.2	20
15	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
16	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
17	Challenges to informed consent for exome sequencing: A bestâ€“worst scaling experiment. Journal of Genetic Counseling, 2019, 28, 1189-1197.	1.6	7
18	Vigabatrin as a Targeted Treatment of GABAB Receptor-Related Epileptic Encephalopathy. Pediatric Neurology, 2019, 99, 82-84.	2.1	2

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19	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
20	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
21	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
22	Mitochondrial DNA Deletions With Low-Level Heteroplasmy in Adult-Onset Myopathy. Journal of Clinical Neuromuscular Disease, 2018, 19, 117-123.	0.7	6
23	FOXG1 syndrome: genotypeâ€“phenotype association in 83 patients with FOXG1 variants. Genetics in Medicine, 2018, 20, 98-108.	2.4	77
24	Successful treatment of choreo-athetotic movements in a patient with an EEF1A2 gene variant. SAGE Open Medical Case Reports, 2018, 6, 2050313X1880762.	0.3	5
25	Clinical wholeâ€exome sequencing results impact medical management. Molecular Genetics & Genomic Medicine, 2018, 6, 1068-1078.	1.2	33
26	ST3GAL5-Related Disorders: A Deficiency in Ganglioside Metabolism and a Genetic Cause of Intellectual Disability and Choreoathetosis. Journal of Child Neurology, 2018, 33, 825-831.	1.4	28
27	Compound Heterozygous Variants in ROBO1 Cause a Neurodevelopmental Disorder With Absence of Transverse Pontine Fibers and Thinning of the Anterior Commissure and Corpus Callosum. Pediatric Neurology, 2017, 70, 70-74.	2.1	16
28	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. American Journal of Human Genetics, 2017, 101, 1013-1020.	6.2	53
29	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
30	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	2.8	36
31	ELP2 is a novel gene implicated in neurodevelopmental disabilities. American Journal of Medical Genetics, Part A, 2015, 167, 1391-1395.	1.2	61
32	A Diagnostic Approach for Cerebral Palsy in the Genomic Era. NeuroMolecular Medicine, 2014, 16, 821-844.	3.4	89
33	Clinical whole exome sequencing in child neurology practice. Annals of Neurology, 2014, 76, 473-483.	5.3	228
34	MEF2C Haploinsufficiency features consistent hyperkinesis, variable epilepsy, and has a role in dorsal and ventral neuronal developmental pathways. Neurogenetics, 2013, 14, 99-111.	1.4	89
35	Development and validation of the Psychological Adaptation Scale (PAS): Use in six studies of adaptation to a health condition or risk. Patient Education and Counseling, 2013, 93, 248-254.	2.2	54
36	Diagnostic Evaluation in Children With Developmental Delay. Clinical Pediatrics, 2012, 51, 1208-1210.	0.8	3

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37	Constitutional Duplication of a Region of Chromosome Yp Encoding AMELY, PRKY, and TBL1Y. Journal of Molecular Diagnostics, 2007, 9, 408-413.	2.8	23
38	RANBP2 and CLTC are involved in ALK rearrangements in inflammatory myofibroblastic tumors. Cancer Genetics and Cytogenetics, 2007, 176, 107-114.	1.0	72