Jill V Hunter

List of Publications by Year
in descending order

58 papers	2,168 citations	257450 24 h-index	254184 43 g-index
58 all docs	58 docs citations	58 times ranked	5263 citing authors

#	Article	IF	Citations
1	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
2	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
3	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	28.9	189
4	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
5	Emerging Imaging Tools for Use with Traumatic Brain Injury Research. Journal of Neurotrauma, 2012, 29, 654-671.	3.4	121
6	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	8.1	95
7	Multi-modal MRI of mild traumatic brain injury. NeuroImage: Clinical, 2015, 7, 87-97.	2.7	82
8	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	6.2	66
9	Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia (MLASA) plus associated with a novel de novo mutation (m.8969G>A) in the mitochondrial encoded ATP6 gene. Molecular Genetics and Metabolism, 2014, 113, 207-212.	1.1	63
10	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	6.2	57
11	Mutations inVRK1Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. JAMA Neurology, 2013, 70, 1491-8.	9.0	54
12	Cortical Thickness in Mild Traumatic Brain Injury. Journal of Neurotrauma, 2016, 33, 1809-1817.	3.4	54
13	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. European Journal of Human Genetics, 2015, 23, 54-60.	2.8	45
14	Functional Connectivity Is Altered in Concussed Adolescent Athletes Despite Medical Clearance to Return to Play: A Preliminary Report. Frontiers in Neurology, 2016, 7, 116.	2.4	45
15	Orthopedic Injured versus Uninjured Comparison Groups for Neuroimaging Research in Mild Traumatic Brain Injury. Journal of Neurotrauma, 2019, 36, 239-249.	3.4	45
16	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	2.4	41
17	Loss of Consciousness Is Related to White Matter Injury in Mild Traumatic Brain Injury. Journal of Neurotrauma, 2016, 33, 2000-2010.	3.4	40
18	Late proton MR spectroscopy in children after traumatic brain injury: correlation with cognitive outcomes. American Journal of Neuroradiology, 2005, 26, 482-8.	2.4	40

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19	The phenotypic spectrum of Xiaâ€Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326.	1.2	34
20	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	7.6	33
21	Pediatric traumatic brain injury: Neuroimaging and neurorehabilitation outcome. NeuroRehabilitation, 2012, 31, 245-260.	1.3	31
22	A preliminary report of cerebral white matter microstructural changes associated with adolescent sports concussion acutely and subacutely using diffusion tensor imaging. Brain Imaging and Behavior, 2018, 12, 962-973.	2.1	29
23	Chronic Maternal Hyperoxygenation and Effect on Cerebral and Placental Vasoregulation and Neurodevelopment in Fetuses with Left Heart Hypoplasia. Fetal Diagnosis and Therapy, 2019, 46, 45-57.	1.4	29
24	Chronic Effects of Boxing: Diffusion Tensor Imaging and Cognitive Findings. Journal of Neurotrauma, 2016, 33, 672-680.	3.4	28
25	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	6.2	24
26	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
27	Cerebral oxygen metabolism during and after therapeutic hypothermia in neonatal hypoxicâ€“ischemic encephalopathy: a feasibility study using magnetic resonance imaging. Pediatric Radiology, 2019, 49, 224-233.	2.0	21
28	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	3.7	20
29	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
30	Increased bone turnover, osteoporosis, progressive tibial bowing, fractures, and scoliosis in a patient with a finalâ€exon SATB2 frameshift mutation. American Journal of Medical Genetics, Part A, 2016, 170, 3028-3032.	1.2	16
31	Volumetric brain magnetic resonance imaging analysis in children with obstructive sleep apnea. International Journal of Pediatric Otorhinolaryngology, 2020, 138, 110369.	1.0	16
32	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
33	Methylphenidate Treatment of Cognitive Dysfunction in Adults After Mild to Moderate Traumatic Brain Injury: Rationale, Efficacy, and Neural Mechanisms. Frontiers in Neurology, 2019, 10, 925.	2.4	15
34	Acute pediatric traumatic brain injury severity predicts long-term verbal memory performance through suppression by white matter integrity on diffusion tensor imaging. Brain Imaging and Behavior, 2020, 14, 1626-1637.	2.1	15
35	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	3.7	15
36	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14

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37	A preliminary investigation of corpus callosum subregion white matter vulnerability and relation to chronic outcome in boxers. Brain Imaging and Behavior, 2020, 14, 772-786.	2.1	13
38	Diffusion Tensor Imaging Indicators of White Matter Injury Are Correlated with a Multimodal Electroencephalography-Based Biomarker in Slow Recovering, Concussed Collegiate Athletes. Journal of Neurotrauma, 2020, 37, 2093-2101.	3.4	13
39	Neurodevelopmental disorder in an Egyptian family with a biallelic <scp>ALKBH8</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 1288-1293.	1.2	13
40	A novel CACNA1A variant in a child with early stroke and intractable epilepsy. Molecular Genetics & Genomic Medicine, 2020, 8, e1383.	1.2	11
41	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460.	2.4	9
42	Persistent Disruption of Brain Connectivity after Sports-Related Concussion in a Female Athlete. Journal of Neurotrauma, 2019, 36, 3164-3171.	3.4	8
43	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83.	7.6	8
44	Two novel bia€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249.	1.2	7
45	<scp>Ela€Hattaba€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540.	2.0	7
46	MRI venous architecture of the thalamus. Journal of the Neurological Sciences, 2016, 370, 88-93.	0.6	6
47	Developmental Alterations in Cortical Organization and Socialization in Adolescents Who Sustained a Traumatic Brain Injury in Early Childhood. Journal of Neurotrauma, 2021, 38, 133-143.	3.4	6
48	Risk of sudden cardiac death in <scp>EXOSC5</scp>a€related disease. American Journal of Medical Genetics, Part A, 2021, 185, 2532-2540.	1.2	6
49	Methionyl-tRNA Formyltransferase (MTFMT) Deficiency Mimicking Acquired Demyelinating Disease. Journal of Child Neurology, 2016, 31, 215-219.	1.4	5
50	The Spatial Relationship of Child Homicides to Community Resources in a Large Metropolitan Area. SAGE Open, 2013, 3, 215824401348313.	1.7	4
51	GNA11 brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis. Neurology: Genetics, 2019, 5, e366.	1.9	4
52	Selective Impairment of Inhibition After TBI in Children. Journal of Clinical and Experimental Neuropsychology, 2004, 26, 589-597.	1.3	4
53	Diffusion Tensor Imaging Correlates of Resilience Following Adolescent Traumatic Brain Injury. Cognitive and Behavioral Neurology, 2021, 34, 259-274.	0.9	4
54	BANNWARTH SYNDROME. Pediatric Infectious Disease Journal, 2021, Publish Ahead of Print, e442-e444.	2.0	3

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55	A Preliminary DTI Tractography Study of Developmental Neuroplasticity 5â€“15 Years After Early Childhood Traumatic Brain Injury. Frontiers in Neurology, 2021, 12, 734055.	2.4	3
56	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	1.2	2
57	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	5.3	2
58	Teaching NeuroImages: Spinal cord infarct due to fibrocartilaginous embolism in an adolescent. Neurology, 2020, 94, e2495-e2496.	1.1	0