

Jill V Hunter

List of Publications by Year in descending order

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Version: 2024-02-01

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	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	7.6	17
2	<i>El-Hattab-Alkuraya</i> syndrome caused by biallelic <i>WDR45B</i> pathogenic variants: Further delineation of the phenotype and genotype. <i>Clinical Genetics</i> , 2022, 101, 530-540.	2.0	7
3	Biallelic Variants in the Ectonucleotidase <i>ENTPD1</i> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. <i>Annals of Neurology</i> , 2022, 92, 304-321.	5.3	2
4	Developmental Alterations in Cortical Organization and Socialization in Adolescents Who Sustained a Traumatic Brain Injury in Early Childhood. <i>Journal of Neurotrauma</i> , 2021, 38, 133-143.	3.4	6
5	<i>MED27</i> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. <i>Annals of Neurology</i> , 2021, 89, 828-833.	5.3	14
6	Neurodevelopmental disorder in an Egyptian family with a biallelic <i>ALKBH8</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1288-1293.	1.2	13
7	Two novel biallelic <i>KDEL2</i> missense variants cause osteogenesis imperfecta with neurodevelopmental features. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2241-2249.	1.2	7
8	A novel de novo intronic variant in <i>ITPR1</i> causes Gillespie syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2315-2324.	1.2	2
9	Biallelic and monoallelic variants in <i>PLXNA1</i> are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. <i>Genetics in Medicine</i> , 2021, 23, 1715-1725.	2.4	22
10	Risk of sudden cardiac death in <i>EXOSC5</i> -related disease. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2532-2540.	1.2	6
11	BANNWARTH SYNDROME. <i>Pediatric Infectious Disease Journal</i> , 2021, Publish Ahead of Print, e442-e444.	2.0	3
12	Biallelic loss-of-function variants in the splicing regulator <i>NSRP1</i> cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. <i>Genetics in Medicine</i> , 2021, 23, 2455-2460.	2.4	9
13	Diffusion Tensor Imaging Correlates of Resilience Following Adolescent Traumatic Brain Injury. <i>Cognitive and Behavioral Neurology</i> , 2021, 34, 259-274.	0.9	4
14	A Preliminary DTI Tractography Study of Developmental Neuroplasticity 5-15 Years After Early Childhood Traumatic Brain Injury. <i>Frontiers in Neurology</i> , 2021, 12, 734055.	2.4	3
15	A preliminary investigation of corpus callosum subregion white matter vulnerability and relation to chronic outcome in boxers. <i>Brain Imaging and Behavior</i> , 2020, 14, 772-786.	2.1	13
16	Acute pediatric traumatic brain injury severity predicts long-term verbal memory performance through suppression by white matter integrity on diffusion tensor imaging. <i>Brain Imaging and Behavior</i> , 2020, 14, 1626-1637.	2.1	15
17	Deficiencies in vesicular transport mediated by <i>TRAPPC4</i> are associated with severe syndromic intellectual disability. <i>Brain</i> , 2020, 143, 112-130.	7.6	33
18	Biallelic in-frame deletion in <i>TRAPPC4</i> in a family with developmental delay and cerebellar atrophy. <i>Brain</i> , 2020, 143, e83-e83.	7.6	8

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19	A novel <i>CACNA1A</i> variant in a child with early stroke and intractable epilepsy. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1383.	1.2	11
20	Volumetric brain magnetic resonance imaging analysis in children with obstructive sleep apnea. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2020, 138, 110369.	1.0	16
21	Teaching NeuroImages: Spinal cord infarct due to fibrocartilaginous embolism in an adolescent. <i>Neurology</i> , 2020, 94, e2495-e2496.	1.1	0
22	Diffusion Tensor Imaging Indicators of White Matter Injury Are Correlated with a Multimodal Electroencephalography-Based Biomarker in Slow Recovering, Concussed Collegiate Athletes. <i>Journal of Neurotrauma</i> , 2020, 37, 2093-2101.	3.4	13
23	Biallelic <i>GRM7</i> variants cause epilepsy, microcephaly, and cerebral atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 610-627.	3.7	15
24	Biallelic loss of function variants in <i>PPP1R21</i> cause a neurodevelopmental syndrome with impaired endocytic function. <i>Human Mutation</i> , 2019, 40, 267-280.	2.5	15
25	Orthopedic Injured versus Uninjured Comparison Groups for Neuroimaging Research in Mild Traumatic Brain Injury. <i>Journal of Neurotrauma</i> , 2019, 36, 239-249.	3.4	45
26	Biallelic <i>CACNA2D2</i> variants in epileptic encephalopathy and cerebellar atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1395-1406.	3.7	20
27	Bi-allelic Pathogenic Variants in <i>TUBGCP2</i> Cause Microcephaly and Lissencephaly Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 1005-1015.	6.2	24
28	Methylphenidate Treatment of Cognitive Dysfunction in Adults After Mild to Moderate Traumatic Brain Injury: Rationale, Efficacy, and Neural Mechanisms. <i>Frontiers in Neurology</i> , 2019, 10, 925.	2.4	15
29	Pathogenic variants in <i>USP7</i> cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. <i>Genetics in Medicine</i> , 2019, 21, 1797-1807.	2.4	41
30	Persistent Disruption of Brain Connectivity after Sports-Related Concussion in a Female Athlete. <i>Journal of Neurotrauma</i> , 2019, 36, 3164-3171.	3.4	8
31	<i>GNA11</i> brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis. <i>Neurology: Genetics</i> , 2019, 5, e366.	1.9	4
32	Chronic Maternal Hyperoxygenation and Effect on Cerebral and Placental Vasoregulation and Neurodevelopment in Fetuses with Left Heart Hypoplasia. <i>Fetal Diagnosis and Therapy</i> , 2019, 46, 45-57.	1.4	29
33	Cerebral oxygen metabolism during and after therapeutic hypothermia in neonatal hypoxic-ischemic encephalopathy: a feasibility study using magnetic resonance imaging. <i>Pediatric Radiology</i> , 2019, 49, 224-233.	2.0	21
34	The phenotypic spectrum of Xia-Gibbs syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1315-1326.	1.2	34
35	A preliminary report of cerebral white matter microstructural changes associated with adolescent sports concussion acutely and subacutely using diffusion tensor imaging. <i>Brain Imaging and Behavior</i> , 2018, 12, 962-973.	2.1	29
36	Loss of Nardilysin, a Mitochondrial Co-chaperone for α -Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. <i>Neuron</i> , 2017, 93, 115-131.	8.1	95

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37	Functional Connectivity Is Altered in Concussed Adolescent Athletes Despite Medical Clearance to Return to Play: A Preliminary Report. <i>Frontiers in Neurology</i> , 2016, 7, 116.	2.4	45
38	MRI venous architecture of the thalamus. <i>Journal of the Neurological Sciences</i> , 2016, 370, 88-93.	0.6	6
39	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	6.2	146
40	Increased bone turnover, osteoporosis, progressive tibial bowing, fractures, and scoliosis in a patient with a finalâ€œexon <i>SATB2</i> frameshift mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3028-3032.	1.2	16
41	Loss of Consciousness Is Related to White Matter Injury in Mild Traumatic Brain Injury. <i>Journal of Neurotrauma</i> , 2016, 33, 2000-2010.	3.4	40
42	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 562-570.	6.2	66
43	Cortical Thickness in Mild Traumatic Brain Injury. <i>Journal of Neurotrauma</i> , 2016, 33, 1809-1817.	3.4	54
44	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. <i>JAMA Psychiatry</i> , 2016, 73, 20.	11.0	195
45	Chronic Effects of Boxing: Diffusion Tensor Imaging and Cognitive Findings. <i>Journal of Neurotrauma</i> , 2016, 33, 672-680.	3.4	28
46	Methionyl-tRNA Formyltransferase (MTFMT) Deficiency Mimicking Acquired Demyelinating Disease. <i>Journal of Child Neurology</i> , 2016, 31, 215-219.	1.4	5
47	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. <i>Neuron</i> , 2015, 88, 499-513.	8.1	258
48	Multi-modal MRI of mild traumatic brain injury. <i>NeuroImage: Clinical</i> , 2015, 7, 87-97.	2.7	82
49	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. <i>European Journal of Human Genetics</i> , 2015, 23, 54-60.	2.8	45
50	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. <i>Cell</i> , 2014, 157, 636-650.	28.9	189
51	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. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 207-212.	1.1	63
52	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. <i>American Journal of Human Genetics</i> , 2014, 94, 784-789.	6.2	57
53	Mutations in<i>VRK1</i> Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. <i>JAMA Neurology</i> , 2013, 70, 1491-8.	9.0	54
54	The Spatial Relationship of Child Homicides to Community Resources in a Large Metropolitan Area. <i>SAGE Open</i> , 2013, 3, 215824401348313.	1.7	4

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55	Emerging Imaging Tools for Use with Traumatic Brain Injury Research. Journal of Neurotrauma, 2012, 29, 654-671.	3.4	121
56	Pediatric traumatic brain injury: Neuroimaging and neurorehabilitation outcome. NeuroRehabilitation, 2012, 31, 245-260.	1.3	31
57	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
58	Selective Impairment of Inhibition After TBI in Children. Journal of Clinical and Experimental Neuropsychology, 2004, 26, 589-597.	1.3	4