

Paul J Lockhart

List of Publications by Year in descending order

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146
papers

9,259
citations

44069

48
h-index

45317

90
g-index

158
all docs

158
docs citations

158
times ranked

13119
citing authors

#	ARTICLE	IF	CITATIONS
1	Isolation of a partial candidate gene for Menkes disease by positional cloning. <i>Nature Genetics</i> , 1993, 3, 20-25.	21.4	688
2	Refining analyses of copy number variation identifies specific genes associated with developmental delay. <i>Nature Genetics</i> , 2014, 46, 1063-1071.	21.4	583
3	Parkin Protects against the Toxicity Associated with Mutant α -Synuclein. <i>Neuron</i> , 2002, 36, 1007-1019.	8.1	542
4	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. <i>Nature Genetics</i> , 2017, 49, 515-526.	21.4	443
5	alpha-synuclein gene haplotypes are associated with Parkinson's disease. <i>Human Molecular Genetics</i> , 2001, 10, 1847-1851.	2.9	314
6	Translation Initiator EIF4G1 Mutations in Familial Parkinson Disease. <i>American Journal of Human Genetics</i> , 2011, 89, 398-406.	6.2	250
7	UCHL1 is a Parkinson's disease susceptibility gene. <i>Annals of Neurology</i> , 2004, 55, 512-521.	5.3	227
8	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with α -Synuclein Pathology. <i>American Journal of Human Genetics</i> , 2014, 95, 729-735.	6.2	207
9	The PARK8 Locus in Autosomal Dominant Parkinsonism: Confirmation of Linkage and Further Delineation of the Disease-Containing Interval. <i>American Journal of Human Genetics</i> , 2004, 74, 11-19.	6.2	195
10	Metalloprotease SPRTN/DVC1 Orchestrates Replication-Coupled DNA-Protein Crosslink Repair. <i>Molecular Cell</i> , 2016, 64, 704-719.	9.7	193
11	Degeneration in Different Parkinsonian Syndromes Relates to Astrocyte Type and Astrocyte Protein Expression. <i>Journal of Neuropathology and Experimental Neurology</i> , 2009, 68, 1073-1083.	1.7	173
12	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. <i>American Journal of Human Genetics</i> , 2019, 105, 151-165.	6.2	170
13	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. <i>Nature Genetics</i> , 2014, 46, 1239-1244.	21.4	165
14	Parkin genetics: one model for Parkinson's disease. <i>Human Molecular Genetics</i> , 2004, 13, 127R-133.	2.9	153
15	Mutations in DARS Cause Hypomyelination with Brain Stem and Spinal Cord Involvement and Leg Spasticity. <i>American Journal of Human Genetics</i> , 2013, 92, 774-780.	6.2	151
16	The Role of GMXCXXC Metal Binding Sites in the Copper-induced Redistribution of the Menkes Protein. <i>Journal of Biological Chemistry</i> , 1999, 274, 11170-11177.	3.4	150
17	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. <i>Acta Neuropathologica</i> , 2016, 131, 847-863.	7.7	143
18	RING finger 1 mutations in Parkin produce altered localization of the protein. <i>Human Molecular Genetics</i> , 2003, 12, 2957-2965.	2.9	138

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19	Profile of families with parkinsonism-predominant spinocerebellar ataxia type 2 (SCA2). <i>Movement Disorders</i> , 2004, 19, 622-629.	3.9	127
20	Quantitative proteomic analysis of mitochondrial proteins: relevance to Lewy body formation and Parkinson's disease. <i>Molecular Brain Research</i> , 2005, 134, 119-138.	2.3	126
21	Human and Mouse Mutations in WDR35 Cause Short-Rib Polydactyly Syndromes Due to Abnormal Ciliogenesis. <i>American Journal of Human Genetics</i> , 2011, 88, 508-515.	6.2	122
22	Mutations in the murine homologue of the Menkes gene in dappled and blotchy mice. <i>Nature Genetics</i> , 1994, 6, 374-378.	21.4	121
23	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator <i>NPRL3</i> . <i>Annals of Neurology</i> , 2016, 79, 132-137.	5.3	116
24	Compound heterozygous <i>FXN</i> mutations and clinical outcome in friedreich ataxia. <i>Annals of Neurology</i> , 2016, 79, 485-495.	5.3	115
25	SCA-2 presenting as parkinsonism in an Alberta family. <i>Neurology</i> , 2002, 59, 1625-1627.	1.1	113
26	Identification of a Novel Gene Linked to Parkin via a Bi-directional Promoter. <i>Journal of Molecular Biology</i> , 2003, 326, 11-19.	4.2	111
27	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	12.8	105
28	SCA2 may present as levodopa-responsive parkinsonism. <i>Movement Disorders</i> , 2003, 18, 425-429.	3.9	99
29	Correction of the Copper Transport Defect of Menkes Patient Fibroblasts by Expression of the Menkes and Wilson ATPases. <i>Journal of Biological Chemistry</i> , 1998, 273, 31375-31380.	3.4	97
30	Functional association of the parkin gene promoter with idiopathic Parkinson's disease. <i>Human Molecular Genetics</i> , 2002, 11, 2787-2792.	2.9	95
31	Familial cortical dysplasia type IIA caused by a germline mutation in <i>DEPDC5</i> . <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 575-580.	3.7	95
32	Detecting Expansions of Tandem Repeats in Cohorts Sequenced with Short-Read Sequencing Data. <i>American Journal of Human Genetics</i> , 2018, 103, 858-873.	6.2	93
33	An open-label trial in Friedreich ataxia suggests clinical benefit with high-dose resveratrol, without effect on frataxin levels. <i>Journal of Neurology</i> , 2015, 262, 1344-1353.	3.6	89
34	Oligomeric α -synuclein inhibits tubulin polymerization. <i>Biochemical and Biophysical Research Communications</i> , 2007, 356, 548-553.	2.1	86
35	Functional analysis and intracellular localization of the human menkes protein (MNK) stably expressed from a cDNA construct in Chinese hamster ovary cells (CHO-K1). <i>Human Molecular Genetics</i> , 1998, 7, 1293-1300.	2.9	84
36	Recent advances in the detection of repeat expansions with short-read next-generation sequencing. <i>F1000Research</i> , 2018, 7, 736.	1.6	84

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37	Molecular basis of the brindled mouse mutant (Mo(br)): a murine model of Menkes disease. <i>Human Molecular Genetics</i> , 1997, 6, 1037-1042.	2.9	82
38	ASK1 inhibition: a therapeutic strategy with multi-system benefits. <i>Journal of Molecular Medicine</i> , 2020, 98, 335-348.	3.9	75
39	DJ-1 mutations are a rare cause of recessively inherited early onset parkinsonism mediated by loss of protein function. <i>Journal of Medical Genetics</i> , 2004, 41, 22e-22.	3.2	74
40	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. <i>Brain</i> , 2016, 139, 2877-2890.	7.6	74
41	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. <i>Genome Biology</i> , 2011, 12, R85.	9.6	72
42	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. <i>Nature Genetics</i> , 2017, 49, 511-514.	21.4	69
43	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in <i>MTOR</i> . <i>Neurology</i> , 2015, 84, 2029-2032.	1.1	64
44	Expression of the Menkes gene homologue in mouse tissues lack of effect of copper on the mRNA levels. <i>FEBS Letters</i> , 1994, 351, 186-190.	2.8	58
45	It's a double knock-out! The quaking mouse is a spontaneous deletion of parkin and parkin co-regulated gene (PACRG). <i>Movement Disorders</i> , 2004, 19, 101-104.	3.9	58
46	The emerging role of Rab GTPases in the pathogenesis of Parkinson's disease. <i>Movement Disorders</i> , 2018, 33, 196-207.	3.9	55
47	Secondarily DEPDC5 mutation is limited to dysmorphic neurons in cortical dysplasia type IIA. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1338-1344.	3.7	55
48	Intracellular localization and loss of copper responsiveness of Mnk, the murine homologue of the Menkes protein, in cells from blotchy (Mo blo) and brindled (Mo br) mouse mutants. <i>Human Molecular Genetics</i> , 1999, 8, 1069-1075.	2.9	54
49	Deletion of the Parkin co-regulated gene causes defects in ependymal ciliary motility and hydrocephalus in the quakingviable mutant mouse. <i>Human Molecular Genetics</i> , 2010, 19, 1593-1602.	2.9	52
50	ALPK3-deficient cardiomyocytes generated from patient-derived induced pluripotent stem cells and mutant human embryonic stem cells display abnormal calcium handling and establish that ALPK3 deficiency underlies familial cardiomyopathy. <i>European Heart Journal</i> , 2016, 37, 2586-2590.	2.2	49
51	Refinement of the PARK3 locus on chromosome 2p13 and the analysis of 14 candidate genes. <i>European Journal of Human Genetics</i> , 2001, 9, 659-666.	2.8	46
52	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. <i>Brain Communications</i> , 2021, 3, fcaa235.	3.3	42
53	<i>DCC</i> mutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. <i>Human Mutation</i> , 2018, 39, 23-39.	2.5	41
54	Prevalence of <i>RFC1</i> -mediated spinocerebellar ataxia in a North American ataxia cohort. <i>Neurology: Genetics</i> , 2020, 6, e440.	1.9	40

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55	<i>ARID1B</i>-mediated disorders: Mutations and possible mechanisms. <i>Intractable and Rare Diseases Research</i> , 2015, 4, 17-23.	0.9	38
56	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 318-325.	3.7	36
57	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. <i>Journal of Human Genetics</i> , 2019, 64, 1173-1186.	2.3	36
58	Parkin Co-regulated Gene (PACRG) is regulated by the ubiquitinâ€“proteasomal system and is present in the pathological features of parkinsonian diseases. <i>Neurobiology of Disease</i> , 2007, 27, 238-247.	4.4	32
59	DEPDC5 and NPRL3 modulate cell size, filopodial outgrowth, and localization of mTOR in neural progenitor cells and neurons. <i>Neurobiology of Disease</i> , 2018, 114, 184-193.	4.4	32
60	Clinical spectrum of the pentanucleotide repeat expansion in the <i>RFC1</i> gene in ataxia syndromes. <i>Neurology</i> , 2020, 95, e2912-e2923.	1.1	32
61	Identification and characterization of the human parkin gene promoter. <i>Journal of Neurochemistry</i> , 2001, 78, 1146-1152.	3.9	31
62	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. <i>Brain</i> , 2017, 140, 2093-2103.	7.6	31
63	<i>ADGRV1</i> is implicated in myoclonic epilepsy. <i>Epilepsia</i> , 2018, 59, 381-388.	5.1	31
64	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. <i>European Journal of Human Genetics</i> , 2014, 22, 741-747.	2.8	30
65	Genetic characterization identifies bottom-of-sulcus dysplasia as an mTORopathy. <i>Neurology</i> , 2020, 95, e2542-e2551.	1.1	30
66	Cell and Gene Therapy for Friedreich Ataxia: Progress to Date. <i>Human Gene Therapy</i> , 2014, 25, 684-693.	2.7	29
67	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. <i>Neurology: Genetics</i> , 2015, 1, e16.	1.9	29
68	Somatic <i>GNAQ</i> mutation in the <i>forme fruste</i> of Sturge-Weber syndrome. <i>Neurology: Genetics</i> , 2018, 4, e236.	1.9	29
69	Parkin Co-Regulated Gene is involved in aggresome formation and autophagy in response to proteasomal impairment. <i>Experimental Cell Research</i> , 2012, 318, 2059-2070.	2.6	28
70	Generation of iPSC lines from peripheral blood mononuclear cells from 5 healthy adults. <i>Stem Cell Research</i> , 2019, 34, 101380.	0.7	28
71	Parental health spillover effects of paediatric rare genetic conditions. <i>Quality of Life Research</i> , 2020, 29, 2445-2454.	3.1	28
72	Lack of mutations in DJ-1 in a cohort of Taiwanese ethnic Chinese with early-onset parkinsonism. <i>Movement Disorders</i> , 2004, 19, 1065-1069.	3.9	27

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73	Functional analysis of a hypomorphic allele shows that MMP14 catalytic activity is the prime determinant of the Winchester syndrome phenotype. <i>Human Molecular Genetics</i> , 2018, 27, 2775-2788.	2.9	25
74	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. <i>American Heart Journal</i> , 2020, 225, 108-119.	2.7	25
75	Spinocerebellar ataxia type 14: study of a family with an exon 5 mutation in the PRKCG gene. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2005, 76, 1720-1722.	1.9	24
76	Genotype-phenotype correlates in Taiwanese patients with early-onset recessive parkinsonism. <i>Movement Disorders</i> , 2009, 24, 104-108.	3.9	24
77	Gradient of brain mosaic <i>RHEB</i> variants causes a continuum of cortical dysplasia. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 485-490.	3.7	24
78	Mutant torsinA interacts with tyrosine hydroxylase in cultured cells. <i>Neuroscience</i> , 2009, 164, 1127-1137.	2.3	23
79	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 914-924.	6.2	23
80	The human sideroflexin 5 (SFXN5) gene: sequence, expression analysis and exclusion as a candidate for PARK3. <i>Gene</i> , 2002, 285, 229-237.	2.2	22
81	Genetic heterogeneity of polymicrogyria: study of 123 patients using deep sequencing. <i>Brain Communications</i> , 2021, 3, fcaa221.	3.3	22
82	DCC Is Required for the Development of Nociceptive Topognosis in Mice and Humans. <i>Cell Reports</i> , 2018, 22, 1105-1114.	6.4	21
83	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. <i>Neuron</i> , 2020, 106, 237-245.e8.	8.1	21
84	Cloning and expression analysis of the sheep ceruloplasmin cDNA. <i>Gene</i> , 1999, 236, 251-257.	2.2	19
85	Identification of the Human Ubiquitin Specific Protease 31 (USP31) Gene: Structure, Sequence and Expression Analysis. <i>DNA Sequence</i> , 2004, 15, 9-14.	0.7	19
86	De novo 325 kb microdeletion in chromosome band 10q25.3 including ATRNL1 in a boy with cognitive impairment, autism and dysmorphic features. <i>European Journal of Medical Genetics</i> , 2010, 53, 337-339.	1.3	19
87	Distribution of Parkinson's disease associated RAB39B in mouse brain tissue. <i>Molecular Brain</i> , 2020, 13, 52.	2.6	19
88	Accurate Determination of Ataxin-2 Polyglutamine Expansion in Patients with Intermediate-Range Repeats. <i>Genetic Testing and Molecular Biomarkers</i> , 2002, 6, 217-220.	1.7	18
89	Heterozygous mutations in <i>HSD17B4</i> cause juvenile peroxisomal D-bifunctional protein deficiency. <i>Neurology: Genetics</i> , 2016, 2, e114.	1.9	18
90	Cerebral hypomyelination associated with biallelic variants of <i>FIG4</i> . <i>Human Mutation</i> , 2019, 40, 619-630.	2.5	18

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91	Polyalanine expansion mutations in the X-linked hypopituitarism gene SOX3 result in aggresome formation and impaired transactivation. <i>Frontiers in Bioscience - Landmark</i> , 2007, 12, 2085.	3.0	18
92	Expression and localization of the Parkin Co-Regulated Gene in mouse CNS suggests a role in ependymal cilia function. <i>Neuroscience Letters</i> , 2009, 460, 97-101.	2.1	17
93	Neuropathology of childhood-onset basal ganglia degeneration caused by mutation of <i>VAC14</i> . <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 859-864.	3.7	17
94	Eukaryotic Expression Vectors That Replicate to Low Copy Number in Bacteria: Transient Expression of the Menkes Protein. <i>Plasmid</i> , 1998, 39, 245-251.	1.4	16
95	Cloning, mapping and expression analysis of the sheep Wilson disease gene homologue. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 2000, 1491, 229-239.	2.4	16
96	Expanding the phenotypic spectrum of ARID1B-mediated disorders and identification of altered cell-cycle dynamics due to ARID1B haploinsufficiency. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 43.	2.7	16
97	Generation and characterisation of a parkin-Pacrg knockout mouse line and a Pacrg knockout mouse line. <i>Scientific Reports</i> , 2018, 8, 7528.	3.3	16
98	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 601-617.	6.2	16
99	Molecular analysis of the Parkin co-regulated gene and association with male infertility. <i>Fertility and Sterility</i> , 2010, 93, 2262-2268.	1.0	15
100	The COMT Val158 allele is associated with impaired delayed-match-to-sample performance in ADHD. <i>Behavioral and Brain Functions</i> , 2012, 8, 25.	3.3	15
101	Multiplication of the α -Synuclein Gene Is Not a Common Disease Mechanism in Lewy Body Disease. <i>Journal of Molecular Neuroscience</i> , 2004, 24, 337-342.	2.3	14
102	Clinical and Neuropathological Features Associated With Loss of RAB39B. <i>Movement Disorders</i> , 2020, 35, 687-693.	3.9	14
103	Cortical Dysplasia and the mTOR Pathway: How the Study of Human Brain Tissue Has Led to Insights into Epileptogenesis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 1344.	4.1	14
104	Identification of the copper chaperone SAH in Ovis aries: expression analysis and in vitro interaction of SAH with ATP7B. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 2000, 1490, 11-20.	2.4	12
105	Biochemical characterization of torsinB. <i>Molecular Brain Research</i> , 2004, 127, 1-9.	2.3	12
106	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a Three-Generation Family Using Short-Read Whole-Genome Sequencing Data. <i>Movement Disorders</i> , 2020, 35, 1675-1679.	3.9	12
107	Regional and cellular localisation of Parkin Co-Regulated Gene in developing and adult mouse brain. <i>Brain Research</i> , 2008, 1201, 177-186.	2.2	11
108	A novel <i>AMPD2</i> mutation outside the AMP deaminase domain causes pontocerebellar hypoplasia type 9. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 820-823.	1.2	11

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109	Genetic Analysis of RAB39B in an Early-Onset Parkinson's Disease Cohort. <i>Frontiers in Neurology</i> , 2020, 11, 523.	2.4	11
110	Callosal agenesis and congenital mirror movements: outcomes associated with <i>DCC</i> mutations. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 758-762.	2.1	11
111	Implementation of ironXS: a study of the acceptability and feasibility of genetic screening for hereditary hemochromatosis in high schools. <i>Clinical Genetics</i> , 2010, 77, 241-248.	2.0	10
112	Severe Leukoencephalopathy with Clinical Recovery Caused by Recessive BOLA3 Mutations. <i>JIMD Reports</i> , 2018, 43, 63-70.	1.5	10
113	Intravenously delivered aminoglycoside antibiotics, tobramycin and amikacin, are not ototoxic in mice. <i>Hearing Research</i> , 2020, 386, 107870.	2.0	10
114	Functional analysis of the sheep Wilson disease protein (sATP7B) in CHO cells. <i>European Journal of Cell Biology</i> , 2001, 80, 349-357.	3.6	9
115	Long Range Regulation of Human FXN Gene Expression. <i>PLoS ONE</i> , 2011, 6, e22001.	2.5	9
116	Identification of a Novel RNF213 Variant in a Family with Heterogeneous Intracerebral Vasculopathy. <i>International Journal of Stroke</i> , 2014, 9, E26-E27.	5.9	9
117	HFE p.C282Y heterozygosity is associated with earlier disease onset in Friedreich ataxia. <i>Movement Disorders</i> , 2014, 29, 940-943.	3.9	9
118	Genetic Analysis of Patients Who Experienced Awareness with Recall while under General Anesthesia. <i>Anesthesiology</i> , 2019, 131, 974-982.	2.5	9
119	Chudley-McCullough Syndrome: A Recognizable Clinical Entity Characterized by Deafness and Typical Brain Malformations. <i>Journal of Child Neurology</i> , 2021, 36, 152-158.	1.4	9
120	Analysis of PARKIN Co-Regulated Gene in a Taiwanese "Ethnic Chinese cohort with early-onset Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2009, 15, 417-421.	2.2	8
121	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. <i>Human Molecular Genetics</i> , 2022, 31, 2307-2316.	2.9	8
122	Heterozygous <i>PNPT1</i> Variants Cause Spinocerebellar Ataxia Type 25. <i>Annals of Neurology</i> , 2022, 92, 122-137.	5.3	8
123	Correction of the copper transport defect of Menkes patient fibroblasts by expression of two forms of the sheep Wilson ATPase. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2002, 1588, 189-194.	3.8	7
124	ironXS: high-school screening for hereditary haemochromatosis is acceptable and feasible. <i>European Journal of Human Genetics</i> , 2012, 20, 505-509.	2.8	7
125	Generation of RAB39B knockout isogenic human embryonic stem cell lines to model RAB39B-mediated Parkinson's disease. <i>Stem Cell Research</i> , 2018, 28, 161-164.	0.7	7
126	Familial early onset Parkinson's disease caused by a homozygous frameshift variant in PARK7: Clinical features and literature update. <i>Parkinsonism and Related Disorders</i> , 2019, 64, 308-311.	2.2	7

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127	Polymicrogyria associated with 17p13.3p13.2 duplication: Case report and review of the literature. <i>European Journal of Medical Genetics</i> , 2020, 63, 103774.	1.3	7
128	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 749-756.	6.2	6
129	CUGC for pontocerebellar hypoplasia type 9 and spastic paraplegia-63. <i>European Journal of Human Genetics</i> , 2019, 27, 161-166.	2.8	5
130	DCC regulates astroglial development essential for telencephalic morphogenesis and corpus callosum formation. <i>ELife</i> , 2021, 10, .	6.0	5
131	Advancing the diagnosis of repeat expansion disorders. <i>Lancet Neurology</i> , The, 2022, 21, 205-207.	10.2	5
132	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. <i>Stem Cell Research</i> , 2019, 39, 101516.	0.7	4
133	Resection of tuber centers only for seizure control in tuberous sclerosis complex. <i>Epilepsy Research</i> , 2021, 171, 106572.	1.6	4
134	Functional Analysis of the Menkes Protein (MNK) Expressed from a cDNA Construct. <i>Advances in Experimental Medicine and Biology</i> , 1999, 448, 67-82.	1.6	4
135	Lack of evidence for association of a parkin promoter polymorphism with early-onset Parkinson's disease in a Chinese population. <i>Parkinsonism and Related Disorders</i> , 2009, 15, 149-152.	2.2	3
136	Rasmussen encephalitis tissue transfer program. <i>Epilepsia</i> , 2016, 57, 1005-1007.	5.1	3
137	Tracing Autism Traits in Large Multiplex Families to Identify Endophenotypes of the Broader Autism Phenotype. <i>International Journal of Molecular Sciences</i> , 2020, 21, 7965.	4.1	3
138	Clonally Focused Public and Private T Cells in Resected Brain Tissue From Surgeries to Treat Children With Intractable Seizures. <i>Frontiers in Immunology</i> , 2021, 12, 664344.	4.8	3
139	ASK1 is a novel molecular target for preventing aminoglycoside-induced hair cell death. <i>Journal of Molecular Medicine</i> , 2022, 100, 797-813.	3.9	3
140	Teaching NeuroImages: Imaging features of DCC-mediated mirror movements and isolated agenesis of the corpus callosum. <i>Neurology</i> , 2018, 91, e886-e887.	1.1	2
141	Expanding the clinical and radiological phenotypes of leukoencephalopathy due to biallelic <i>HMBS</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2941-2950.	1.2	2
142	A family study implicates <i>GBE1</i> in the etiology of autism spectrum disorder. <i>Human Mutation</i> , 2022, 43, 16-29.	2.5	2
143	Identification and validation of control cell lines for accurate parkin dosage analysis. <i>Journal of Neuroscience Methods</i> , 2009, 176, 68-71.	2.5	1
144	Generation of four iPSC lines from Neurofibromatosis Type 1 patients. <i>Stem Cell Research</i> , 2020, 49, 102013.	0.7	1

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145	Identification of a Novel Gene Linked to Parkin via a Bidirectional Promoter. Annals of the New York Academy of Sciences, 2003, 991, 311-314.	3.8	0
146	Clinical seizure manifestations in the absence of synaptic connections. Epileptic Disorders, 2021, 23, 167-172.	1.3	0