Paul J Lockhart

List of Publications by Year
in descending order

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	A family study implicates GBE1 in the etiology of autism spectrum disorder. Human Mutation, 2022, 43, 16-29.	2.5	2
2	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	2.9	8
3	Cortical Dysplasia and the mTOR Pathway: How the Study of Human Brain Tissue Has Led to Insights into Epileptogenesis. International Journal of Molecular Sciences, 2022, 23, 1344.	4.1	14
4	Advancing the diagnosis of repeat expansion disorders. Lancet Neurology, The, 2022, 21, 205-207.	10.2	5
5	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
6	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	5.3	8
7	ASK1 is a novel molecular target for preventing aminoglycoside-induced hair cell death. Journal of Molecular Medicine, 2022, 100, 797-813.	3.9	3
8	Chudley-McCullough Syndrome: A Recognizable Clinical Entity Characterized by Deafness and Typical Brain Malformations. Journal of Child Neurology, 2021, 36, 152-158.	1.4	9
9	Gradient of brain mosaic RHEB variants causes a continuum of cortical dysplasia. Annals of Clinical and Translational Neurology, 2021, 8, 485-490.	3.7	24
10	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. Brain Communications, 2021, 3, fcaa235.	3.3	42
11	Clinical seizure manifestations in the absence of synaptic connections. Epileptic Disorders, 2021, 23, 167-172.	1.3	0
12	Resection of tuber centers only for seizure control in tuberous sclerosis complex. Epilepsy Research, 2021, 171, 106572.	1.6	4
13	Clonally Focused Public and Private T Cells in Resected Brain Tissue From Surgeries to Treat Children With Intractable Seizures. Frontiers in Immunology, 2021, 12, 664344.	4.8	3
14	DCC regulates astroglial development essential for telencephalic morphogenesis and corpus callosum formation. ELife, 2021, 10, .	6.0	5
15	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	6.2	6
16	Expanding the clinical and radiological phenotypes of leukoencephalopathy due to biallelic <scp>HMBS</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 2941-2950.	1.2	2
17	Genetic heterogeneity of polymicrogyria: study of 123 patients using deep sequencing. Brain Communications, 2021, 3, fcaa221.	3.3	22
18	Polymicrogyria associated with 17p13.3p13.2 duplication: Case report and review of the literature. European Journal of Medical Genetics, 2020, 63, 103774.	1.3	7

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19	Intravenously delivered aminoglycoside antibiotics, tobramycin and amikacin, are not ototoxic in mice. Hearing Research, 2020, 386, 107870.	2.0	10
20	Generation of four iPSC lines from Neurofibromatosis Type 1 patients. Stem Cell Research, 2020, 49, 102013.	0.7	1
21	Clinical spectrum of the pentanucleotide repeat expansion in the RFC1 gene in ataxia syndromes. Neurology, 2020, 95, e2912-e2923.	1.1	32
22	Genetic characterization identifies bottom-of-sulcus dysplasia as an mTORopathy. Neurology, 2020, 95, e2542-e2551.	1.1	30
23	Tracing Autism Traits in Large Multiplex Families to Identify Endophenotypes of the Broader Autism Phenotype. International Journal of Molecular Sciences, 2020, 21, 7965.	4.1	3
24	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	2.7	25
25	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a <scp>Threeâ€Generation</scp> Family Using <scp>Shortâ€Read Wholeâ€Genome</scp> Sequencing Data. Movement Disorders, 2020, 35, 1675-1679.	3.9	12
26	Prevalence of RFC1 -mediated spinocerebellar ataxia in a North American ataxia cohort. Neurology: Genetics, 2020, 6, e440.	1.9	40
27	Distribution of Parkinsonâ€™s disease associated RAB39B in mouse brain tissue. Molecular Brain, 2020, 13, 52.	2.6	19
28	Genetic Analysis of RAB39B in an Early-Onset Parkinson's Disease Cohort. Frontiers in Neurology, 2020, 11, 523.	2.4	11
29	ASK1 inhibition: a therapeutic strategy with multi-system benefits. Journal of Molecular Medicine, 2020, 98, 335-348.	3.9	75
30	Callosal agenesis and congenital mirror movements: outcomes associated with DCC mutations. Developmental Medicine and Child Neurology, 2020, 62, 758-762.	2.1	11
31	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. Neuron, 2020, 106, 237-245.e8.	8.1	21
32	Clinical and Neuropathological Features Associated With Loss of RAB39B. Movement Disorders, 2020, 35, 687-693.	3.9	14
33	Parental health spillover effects of paediatric rare genetic conditions. Quality of Life Research, 2020, 29, 2445-2454.	3.1	28
34	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
35	CUGC for pontocerebellar hypoplasia type 9 and spastic paraplegia-63. European Journal of Human Genetics, 2019, 27, 161-166.	2.8	5
36	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. Stem Cell Research, 2019, 39, 101516.	0.7	4

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37	Secondâ€hit DEPDC5 mutation is limited to dysmorphic neurons in cortical dysplasia type IIA. Annals of Clinical and Translational Neurology, 2019, 6, 1338-1344.	3.7	55
38	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
39	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. American Journal of Human Genetics, 2019, 105, 151-165.	6.2	170
40	Familial early onset Parkinson's disease caused by a homozygous frameshift variant in PARK7: Clinical features and literature update. Parkinsonism and Related Disorders, 2019, 64, 308-311.	2.2	7
41	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
42	Cerebral hypomyelination associated with biallelic variants of FIG4. Human Mutation, 2019, 40, 619-630.	2.5	18
43	Genetic Analysis of Patients Who Experienced Awareness with Recall while under General Anesthesia. Anesthesiology, 2019, 131, 974-982.	2.5	9
44	Generation of iPSC lines from peripheral blood mononuclear cells from 5 healthy adults. Stem Cell Research, 2019, 34, 101380.	0.7	28
45	DEPDC5 and NPRL3 modulate cell size, filopodial outgrowth, and localization of mTOR in neural progenitor cells and neurons. Neurobiology of Disease, 2018, 114, 184-193.	4.4	32
46	Generation of RAB39B knockout isogenic human embryonic stem cell lines to model RAB39B-mediated Parkinson's disease. Stem Cell Research, 2018, 28, 161-164.	0.7	7
47	Severe Leukoencephalopathy with Clinical Recovery Caused by Recessive BOLA3 Mutations. JIMD Reports, 2018, 43, 63-70.	1.5	10
48	DCC Is Required for the Development of Nociceptive Topognosis in Mice and Humans. Cell Reports, 2018, 22, 1105-1114.	6.4	21
49	ADGRV1 is implicated in myoclonic epilepsy. Epilepsia, 2018, 59, 381-388.	5.1	31
50	The emerging role of Rab GTPases in the pathogenesis of Parkinson's disease. Movement Disorders, 2018, 33, 196-207.	3.9	55
51	DCCmutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. Human Mutation, 2018, 39, 23-39.	2.5	41
52	Detecting Expansions of Tandem Repeats in Cohorts Sequenced with Short-Read Sequencing Data. American Journal of Human Genetics, 2018, 103, 858-873.	6.2	93
53	Teaching NeuroImages: Imaging features of DCC-mediated mirror movements and isolated agenesis of the corpus callosum. Neurology, 2018, 91, e886-e887.	1.1	2
54	Generation and characterisation of a parkin-Pacrg knockout mouse line and a Pacrg knockout mouse line. Scientific Reports, 2018, 8, 7528.	3.3	16

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55	Somatic GNAQ mutation in the forme fruste of Sturge-Weber syndrome. Neurology: Genetics, 2018, 4, e236.	1.9	29
56	Functional analysis of a hypomorphic allele shows that MMP14 catalytic activity is the prime determinant of the Winchester syndrome phenotype. Human Molecular Genetics, 2018, 27, 2775-2788.	2.9	25
57	Recent advances in the detection of repeat expansions with short-read next-generation sequencing. F1000Research, 2018, 7, 736.	1.6	84
58	A novel AMPD2 mutation outside the AMP deaminase domain causes pontocerebellar hypoplasia type 9. American Journal of Medical Genetics, Part A, 2017, 173, 820-823.	1.2	11
59	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	21.4	69
60	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
61	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	3.7	36
62	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	7.6	31
63	Neuropathology of childhoodâ€onset basal ganglia degeneration caused by mutation of VAC14. Annals of Clinical and Translational Neurology, 2017, 4, 859-864.	3.7	17
64	Rasmussen encephalitis tissue transfer program. Epilepsia, 2016, 57, 1005-1007.	5.1	3
65	Heterozygous mutations in HSD17B4 cause juvenile peroxisomal D-bifunctional protein deficiency. Neurology: Genetics, 2016, 2, e114.	1.9	18
66	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. European Heart Journal, 2016, 37, 2586-2590.	2.2	49
67	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator NPRL3. Annals of Neurology, 2016, 79, 132-137.	5.3	116
68	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890.	7.6	74
69	Metalloprotease SPRTN/DVC1 Orchestrates Replication-Coupled DNA-Protein Crosslink Repair. Molecular Cell, 2016, 64, 704-719.	9.7	193
70	Compound heterozygous FXN mutations and clinical outcome in friedreich ataxia. Annals of Neurology, 2016, 79, 485-495.	5.3	115
71	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. Acta Neuropathologica, 2016, 131, 847-863.	7.7	143
72	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. Neurology: Genetics, 2015, 1, e16.	1.9	29

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73	<i>ARID1B</i>-mediated disorders: Mutations and possible mechanisms. Intractable and Rare Diseases Research, 2015, 4, 17-23.	0.9	38
74	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in MTOR. Neurology, 2015, 84, 2029-2032.	1.1	64
75	Familial cortical dysplasia type <scp>IIA</scp> caused by a germline mutation in <scp>DEPDC</scp>5. Annals of Clinical and Translational Neurology, 2015, 2, 575-580.	3.7	95
76	An open-label trial in Friedreich ataxia suggests clinical benefit with high-dose resveratrol, without effect on frataxin levels. Journal of Neurology, 2015, 262, 1344-1353.	3.6	89
77	Identification of a Novel RNF213 Variant in a Family with Heterogeneous Intracerebral Vasculopathy. International Journal of Stroke, 2014, 9, E26-E27.	5.9	9
78	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with Î±-Synuclein Pathology. American Journal of Human Genetics, 2014, 95, 729-735.	6.2	207
79	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. European Journal of Human Genetics, 2014, 22, 741-747.	2.8	30
80	HFE p.C282Y heterozygosity is associated with earlier disease onset in Friedreich ataxia. Movement Disorders, 2014, 29, 940-943.	3.9	9
81	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
82	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nature Genetics, 2014, 46, 1239-1244.	21.4	165
83	Expanding the phenotypic spectrum of ARID1B-mediated disorders and identification of altered cell-cycle dynamics due to ARID1B haploinsufficiency. Orphanet Journal of Rare Diseases, 2014, 9, 43.	2.7	16
84	Cell and Gene Therapy for Friedreich Ataxia: Progress to Date. Human Gene Therapy, 2014, 25, 684-693.	2.7	29
85	Mutations in DARS Cause Hypomyelination with Brain Stem and Spinal Cord Involvement and Leg Spasticity. American Journal of Human Genetics, 2013, 92, 774-780.	6.2	151
86	ironXS: high-school screening for hereditary haemochromatosis is acceptable and feasible. European Journal of Human Genetics, 2012, 20, 505-509.	2.8	7
87	Parkin Co-Regulated Gene is involved in aggresome formation and autophagy in response to proteasomal impairment. Experimental Cell Research, 2012, 318, 2059-2070.	2.6	28
88	The COMT Val158 allele is associated with impaired delayed-match-to-sample performance in ADHD. Behavioral and Brain Functions, 2012, 8, 25.	3.3	15
89	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. Genome Biology, 2011, 12, R85.	9.6	72
90	Human and Mouse Mutations in WDR35 Cause Short-Rib Polydactyly Syndromes Due to Abnormal Ciliogenesis. American Journal of Human Genetics, 2011, 88, 508-515.	6.2	122

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91	Long Range Regulation of Human FXN Gene Expression. PLoS ONE, 2011, 6, e22001.	2.5	9
92	Translation Initiator EIF4G1 Mutations in Familial Parkinson Disease. American Journal of Human Genetics, 2011, 89, 398-406.	6.2	250
93	Implementation of ironXS: a study of the acceptability and feasibility of genetic screening for hereditary hemochromatosis in high schools. Clinical Genetics, 2010, 77, 241-248.	2.0	10
94	Deletion of the Parkin co-regulated gene causes defects in ependymal ciliary motility and hydrocephalus in the quakingviable mutant mouse. Human Molecular Genetics, 2010, 19, 1593-1602.	2.9	52
95	De novo 325 kb microdeletion in chromosome band 10q25.3 including ATRNL1 in a boy with cognitive impairment, autism and dysmorphic features. European Journal of Medical Genetics, 2010, 53, 337-339.	1.3	19
96	Molecular analysis of the PArkin co-regulated gene and association with male infertility. Fertility and Sterility, 2010, 93, 2262-2268.	1.0	15
97	Genotypeâ€“phenotype correlates in Taiwanese patients with earlyâ€onset recessive parkinsonism. Movement Disorders, 2009, 24, 104-108.	3.9	24
98	Identification and validation of control cell lines for accurate parkin dosage analysis. Journal of Neuroscience Methods, 2009, 176, 68-71.	2.5	1
99	Mutant torsinA interacts with tyrosine hydroxylase in cultured cells. Neuroscience, 2009, 164, 1127-1137.	2.3	23
100	Lack of evidence for association of a parkin promoter polymorphism with early-onset Parkinson's disease in a Chinese population. Parkinsonism and Related Disorders, 2009, 15, 149-152.	2.2	3
101	Analysis of PArkin Co-Regulated Gene in a Taiwaneseâ€“Ethnic Chinese cohort with early-onset Parkinson's disease. Parkinsonism and Related Disorders, 2009, 15, 417-421.	2.2	8
102	Expression and localization of the Parkin Co-Regulated Gene in mouse CNS suggests a role in ependymal cilia function. Neuroscience Letters, 2009, 460, 97-101.	2.1	17
103	Degeneration in Different Parkinsonian Syndromes Relates to Astrocyte Type and Astrocyte Protein Expression. Journal of Neuropathology and Experimental Neurology, 2009, 68, 1073-1083.	1.7	173
104	Regional and cellular localisation of Parkin Co-Regulated Gene in developing and adult mouse brain. Brain Research, 2008, 1201, 177-186.	2.2	11
105	Oligomeric Î±-synuclein inhibits tubulin polymerization. Biochemical and Biophysical Research Communications, 2007, 356, 548-553.	2.1	86
106	Parkin Co-regulated Gene (PACRG) is regulated by the ubiquitinâ€“proteasomal system and is present in the pathological features of parkinsonian diseases. Neurobiology of Disease, 2007, 27, 238-247.	4.4	32
107	Polyalanine expansion mutations in the X-linked hypopituitarism gene SOX3 result in aggresome formation and impaired transactivation. Frontiers in Bioscience - Landmark, 2007, 12, 2085.	3.0	18
108	Spinocerebellar ataxia type 14: study of a family with an exon 5 mutation in the PRKCG gene. Journal of Neurology, Neurosurgery and Psychiatry, 2005, 76, 1720-1722.	1.9	24

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109	Quantitative proteomic analysis of mitochondrial proteins: relevance to Lewy body formation and Parkinson's disease. Molecular Brain Research, 2005, 134, 119-138.	2.3	126
110	Parkin genetics: one model for Parkinson's disease. Human Molecular Genetics, 2004, 13, 127R-133.	2.9	153
111	DJ-1 mutations are a rare cause of recessively inherited early onset parkinsonism mediated by loss of protein function. Journal of Medical Genetics, 2004, 41, 22e-22.	3.2	74
112	Multiplication of the Î±-Synuclein Gene Is Not a Common Disease Mechanism in Lewy Body Disease. Journal of Molecular Neuroscience, 2004, 24, 337-342.	2.3	14
113	UCHL1 is a Parkinson's disease susceptibility gene. Annals of Neurology, 2004, 55, 512-521.	5.3	227
114	It's a double knock-out! The quaking mouse is a spontaneous deletion of parkin and parkin co-regulated gene (PACRG). Movement Disorders, 2004, 19, 101-104.	3.9	58
115	Profile of families with parkinsonism-predominant spinocerebellar ataxia type 2 (SCA2). Movement Disorders, 2004, 19, 622-629.	3.9	127
116	Lack of mutations in DJ-1 in a cohort of Taiwanese ethnic Chinese with early-onset parkinsonism. Movement Disorders, 2004, 19, 1065-1069.	3.9	27
117	Identification of the Human Ubiquitin Specific Protease 31 (USP31) Gene: Structure, Sequence and Expression Analysis. DNA Sequence, 2004, 15, 9-14.	0.7	19
118	Biochemical characterization of torsinB. Molecular Brain Research, 2004, 127, 1-9.	2.3	12
119	The PARK8 Locus in Autosomal Dominant Parkinsonism: Confirmation of Linkage and Further Delineation of the Disease-Containing Interval. American Journal of Human Genetics, 2004, 74, 11-19.	6.2	195
120	SCA2 may present as levodopa-responsive parkinsonism. Movement Disorders, 2003, 18, 425-429.	3.9	99
121	Identification of a Novel Gene Linked to Parkin via a Bi-directional Promoter. Journal of Molecular Biology, 2003, 326, 11-19.	4.2	111
122	RING finger 1 mutations in Parkin produce altered localization of the protein. Human Molecular Genetics, 2003, 12, 2957-2965.	2.9	138
123	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
124	Accurate Determination of Ataxin-2 Polyglutamine Expansion in Patients with Intermediate-Range Repeats. Genetic Testing and Molecular Biomarkers, 2002, 6, 217-220.	1.7	18
125	SCA-2 presenting as parkinsonism in an Alberta family. Neurology, 2002, 59, 1625-1627.	1.1	113
126	Functional association of the parkin gene promoter with idiopathic Parkinson's disease. Human Molecular Genetics, 2002, 11, 2787-2792.	2.9	95

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127	Correction of the copper transport defect of Menkes patient fibroblasts by expression of two forms of the sheep Wilson ATPase. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 189-194.	3.8	7
128	Parkin Protects against the Toxicity Associated with Mutant Î±-Synuclein. Neuron, 2002, 36, 1007-1019.	8.1	542
129	The human sideroflexin 5 (SFXN5) gene: sequence, expression analysis and exclusion as a candidate for PARK3. Gene, 2002, 285, 229-237.	2.2	22
130	Identification and characterization of the human parkin gene promoter. Journal of Neurochemistry, 2001, 78, 1146-1152.	3.9	31
131	Refinement of the PARK3 locus on chromosome 2p13 and the analysis of 14 candidate genes. European Journal of Human Genetics, 2001, 9, 659-666.	2.8	46
132	Functional analysis of the sheep Wilson disease protein (sATP7B) in CHO cells. European Journal of Cell Biology, 2001, 80, 349-357.	3.6	9
133	alpha-synuclein gene haplotypes are associated with Parkinson's disease. Human Molecular Genetics, 2001, 10, 1847-1851.	2.9	314
134	Cloning, mapping and expression analysis of the sheep Wilson disease gene homologue. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2000, 1491, 229-239.	2.4	16
135	Identification of the copper chaperone SAH in Ovis aries: expression analysis and in vitro interaction of SAH with ATP7B. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2000, 1490, 11-20.	2.4	12
136	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. Human Molecular Genetics, 1999, 8, 1069-1075.	2.9	54
137	The Role of GMXCXXC Metal Binding Sites in the Copper-induced Redistribution of the Menkes Protein. Journal of Biological Chemistry, 1999, 274, 11170-11177.	3.4	150
138	Cloning and expression analysis of the sheep ceruloplasmin cDNA. Gene, 1999, 236, 251-257.	2.2	19
139	Functional Analysis of the Menkes Protein (MNK) Expressed from a cDNA Construct. Advances in Experimental Medicine and Biology, 1999, 448, 67-82.	1.6	4
140	Eukaryotic Expression Vectors That Replicate to Low Copy Number in Bacteria: Transient Expression of the Menkes Protein. Plasmid, 1998, 39, 245-251.	1.4	16
141	Correction of the Copper Transport Defect of Menkes Patient Fibroblasts by Expression of the Menkes and Wilson ATPases. Journal of Biological Chemistry, 1998, 273, 31375-31380.	3.4	97
142	Functional analysis and intracellular localization of the human menkes protein (MNK) stably expressed from a cDNA construct in Chinese hamster ovary cells (CHO-K1). Human Molecular Genetics, 1998, 7, 1293-1300.	2.9	84
143	Molecular basis of the brindled mouse mutant (Mo(br)): a murine model of Menkes disease. Human Molecular Genetics, 1997, 6, 1037-1042.	2.9	82
144	Mutations in the murine homologue of the Menkes gene in dappled and blotchy mice. Nature Genetics, 1994, 6, 374-378.	21.4	121

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145	Expression of the Menkes gene homologue in mouse tissues lack of effect of copper on the mRNA levels. FEBS Letters, 1994, 351, 186-190.	2.8	58
146	Isolation of a partial candidate gene for Menkes disease by positional cloning. Nature Genetics, 1993, 3, 20-25.	21.4	688