Mark A Corbett

List of Publications by Year
in descending order

80 papers	7,126 citations	76326 40 h-index	62596 80 g-index
83 all docs	83 docs citations	83 times ranked	11035 citing authors

#	Article	IF	Citations
1	Oligonucleotide correction of an intronic TIMMDC1 variant in cells of patients with severe neurodegenerative disorder. Npj Genomic Medicine, 2022, 7, 9.	3.8	8
2	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. Neurology: Genetics, 2022, 8, e652.	1.9	14
3	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.1	13
4	Transgenic mice with an R342X mutation in Phf6 display clinical features of BÃ¶rjesonâ€“Forssmanâ€“Lehmann Syndrome. Human Molecular Genetics, 2021, 30, 575-594.	2.9	5
5	A 127â€‰kb truncating deletion of PGRMC1 is a novel cause of X-linked isolated paediatric cataract. European Journal of Human Genetics, 2021, 29, 1206-1215.	2.8	4
6	Different types of diseaseâ€causing noncoding variants revealed by genomic and gene expression analyses in families with Xâ€linked intellectual disability. Human Mutation, 2021, 42, 835-847.	2.5	0
7	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	2.5	1
8	Yield of clinically reportable genetic variants in unselected cerebral palsy by whole genome sequencing. Npj Genomic Medicine, 2021, 6, 74.	3.8	16
9	Differential gene expression analysis of corneal endothelium indicates involvement of phagocytic activity in Fuchsâ€™ endothelial corneal dystrophy. Experimental Eye Research, 2021, 210, 108692.	2.6	3
10	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
11	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
12	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	21.4	96
13	A synonymous UPF3B variant causing a speech disorder implicates NMD as a regulator of neurodevelopmental disorder gene networks. Human Molecular Genetics, 2020, 29, 2568-2578.	2.9	9
14	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	3.8	17
15	Definition and diagnosis of cerebral palsy in genetic studies: a systematic review. Developmental Medicine and Child Neurology, 2020, 62, 1024-1030.	2.1	16
16	Familial adult myoclonic epilepsy type 1 SAMD12 TTTCA repeat expansion arose 17,000 years ago and is present in Sri Lankan and Indian families. European Journal of Human Genetics, 2020, 28, 973-978.	2.8	23
17	Chromatin-Binding Protein PHF6 Regulates Activity-Dependent Transcriptional Networks to Promote Hunger Response. Cell Reports, 2020, 30, 3717-3728.e6.	6.4	6
18	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99

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19	Reduced expression of apolipoprotein E and immunoglobulin heavy constant gamma 1 proteins in Fuchs endothelial corneal dystrophy. Clinical and Experimental Ophthalmology, 2019, 47, 1028-1042.	2.6	6
20	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. Journal of Child Neurology, 2019, 34, 472-476.	1.4	82
21	PHF6 regulates hematopoietic stem and progenitor cells and its loss synergizes with expression of TLX3 to cause leukemia. Blood, 2019, 133, 1729-1741.	1.4	40
22	Targeted resequencing identifies genes with recurrent variation in cerebral palsy. Npj Genomic Medicine, 2019, 4, 27.	3.8	22
23	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	12.8	111
24	A recurrent missense variant inSLC9A7causes nonsyndromic X-linked intellectual disability with alteration of Golgi acidification and aberrant glycosylation. Human Molecular Genetics, 2019, 28, 598-614.	2.9	25
25	Analysis of 182 cerebral palsy transcriptomes points to dysregulation of trophic signalling pathways and overlap with autism. Translational Psychiatry, 2018, 8, 88.	4.8	22
26	The emerging genetic landscape of cerebral palsy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 331-342.	1.8	23
27	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
28	Pathogenic copy number variants that affect gene expression contribute to genomic burden in cerebral palsy. Npj Genomic Medicine, 2018, 3, 33.	3.8	31
29	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
30	PCDH19 regulation of neural progenitor cell differentiation suggests asynchrony of neurogenesis as a mechanism contributing to PCDH19 Girls Clustering Epilepsy. Neurobiology of Disease, 2018, 116, 106-119.	4.4	39
31	Large deletions induced by Cas9 cleavage. Nature, 2018, 560, E8-E9.	27.8	269
32	Protocadherin 19 (PCDH19) interacts with paraspeckle protein NONO to co-regulate gene expression with estrogen receptor alpha (ERÎ±). Human Molecular Genetics, 2017, 26, 2042-2052.	2.9	28
33	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. European Journal of Medical Genetics, 2017, 60, 437-443.	1.3	10
34	Variant in the X-chromosome spliceosomal gene GPKOW causes male-lethal microcephaly with intrauterine growth restriction. European Journal of Human Genetics, 2017, 25, 1078-1082.	2.8	10
35	A ubiquitin-dependent signalling axis specific for ALKBH-mediated DNA dealkylation repair. Nature, 2017, 551, 389-393.	27.8	83
36	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	3.8	29

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37	HUWE1 mutations in Juberg-Marsidi and Brooks syndromes: the results of an X-chromosome exome sequencing study. BMJ Open, 2016, 6, e009537.	1.9	39
38	A non-coding variant in the 5Ê¹ UTR of DLG3 attenuates protein translation to cause non-syndromic intellectual disability. European Journal of Human Genetics, 2016, 24, 1612-1616.	2.8	10
39	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.1	71
40	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	6.2	247
41	Paternal under-nutrition programs metabolic syndrome in offspring which can be reversed by antioxidant/vitamin food fortification in fathers. Scientific Reports, 2016, 6, 27010.	3.3	56
42	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
43	A mutation in COL4A2 causes autosomal dominant porencephaly with cataracts. American Journal of Medical Genetics, Part A, 2016, 170, 1059-1063.	1.2	17
44	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	7.9	243
45	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.1	23
46	Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204.	2.5	161
47	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
48	Whole-exome sequencing points to considerable genetic heterogeneity of cerebral palsy. Molecular Psychiatry, 2015, 20, 176-182.	7.9	178
49	A novel X-linked trichothiodystrophy associated with a nonsense mutation in RNF113A. Journal of Medical Genetics, 2015, 52, 269-274.	3.2	302
50	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	2.9	25
51	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	6.2	82
52	Developmental disorders: deciphering exomes on a grand scale. Lancet, The, 2015, 385, 1266-1267.	13.7	3
53	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
54	Interchromosomal Insertional Translocation at Xq26.3 AltersSOX3Expression in an Individual With XX Male Sex Reversal. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E815-E820.	3.6	46

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55	shRNA Off-Target Effects In Vivo: Impaired Endogenous siRNA Expression and Spermatogenic Defects. PLoS ONE, 2015, 10, e0118549.	2.5	11
56	Paternal obesity initiates metabolic disturbances in two generations of mice with incomplete penetrance to the F₂generation and alters the transcriptional profile of testis and sperm microRNA content. FASEB Journal, 2013, 27, 4226-4243.	0.5	486
57	De novo intragenic deletion of the autism susceptibility candidate 2 (AUTS2) gene in a patient with developmental delay: A case report and literature review. American Journal of Medical Genetics, Part A, 2013, 161, 1508-1512.	1.2	33
58	TBC1D24 mutation associated with focal epilepsy, cognitive impairment and a distinctive cerebro-cerebellar malformation. Epilepsy Research, 2013, 105, 240-244.	1.6	28
59	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	21.4	301
60	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	7.6	129
61	Expanding the molecular basis and phenotypic spectrum of X-linked Joubert syndrome associated with OFD1 mutations. European Journal of Human Genetics, 2012, 20, 806-809.	2.8	52
62	Copy-Number Gains of HUWE1 Due to Replication- and Recombination-Based Rearrangements. American Journal of Human Genetics, 2012, 91, 252-264.	6.2	71
63	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	6.2	89
64	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. Molecular Psychiatry, 2012, 17, 1103-1115.	7.9	97
65	PRRT2 Mutations Cause Benign Familial Infantile Epilepsy and Infantile Convulsions with Choreoathetosis Syndrome. American Journal of Human Genetics, 2012, 90, 152-160.	6.2	234
66	Identification of a MicroRNA that Activates Gene Expression by Repressing Nonsense-Mediated RNA Decay. Molecular Cell, 2011, 42, 500-510.	9.7	267
67	A Mutation in the Golgi Qb-SNARE Gene GOSR2 Causes Progressive Myoclonus Epilepsy with Early Ataxia. American Journal of Human Genetics, 2011, 88, 657-663.	6.2	166
68	A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	6.2	111
69	CASK mutations are frequent in males and cause X-linked nystagmus and variable XLMR phenotypes. European Journal of Human Genetics, 2010, 18, 544-552.	2.8	105
70	Great expectations: using massively parallel sequencing to solve inherited disorders. Expert Review of Molecular Diagnostics, 2010, 10, 833-836.	3.1	1
71	The genetic landscape of intellectual disability arising from chromosome X. Trends in Genetics, 2009, 25, 308-316.	6.7	190
72	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	21.4	528

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73	Submicroscopic Duplications of the Hydroxysteroid Dehydrogenase HSD17B10 and the E3 Ubiquitin Ligase HUWE1 Are Associated with Mental Retardation. American Journal of Human Genetics, 2008, 82, 432-443.	6.2	187
74	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	21.4	397
75	Mutations in the BRWD3 Gene Cause X-Linked Mental Retardation Associated with Macrocephaly. American Journal of Human Genetics, 2007, 81, 367-374.	6.2	85
76	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. Nature Genetics, 2007, 39, 1127-1133.	21.4	228
77	Protein and gene expression analysis of Phf6, the gene mutated in the BÃ¶rjesonâ€“Forssmanâ€“Lehmann Syndrome of intellectual disability and obesity. Gene Expression Patterns, 2007, 7, 858-871.	0.8	45
78	Skeletal muscle repair in a mouse model of nemaline myopathy. Human Molecular Genetics, 2006, 15, 2603-2612.	2.9	44
79	An ?tropomyosin mutation alters dimer preference in nemaline myopathy. Annals of Neurology, 2005, 57, 42-49.	5.3	62
80	A mutation in alpha-tropomyosinslow affects muscle strength, maturation and hypertrophy in a mouse model for nemaline myopathy. Human Molecular Genetics, 2001, 10, 317-328.	2.9	85