

Mark A Corbett

List of Publications by Year in descending order

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Version: 2024-02-01

80
papers

7,126
citations

76322

40
h-index

62593

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 <i>SLC32A1</i> 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 <i>Phf6</i> display clinical features of Bâ€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 <i>PCDH19</i> missense 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 <i>UPF3B</i> 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 TTCA 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. <i>Clinical and Experimental Ophthalmology</i> , 2019, 47, 1028-1042.	2.6	6
20	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. <i>Journal of Child Neurology</i> , 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. <i>Blood</i> , 2019, 133, 1729-1741.	1.4	40
22	Targeted resequencing identifies genes with recurrent variation in cerebral palsy. <i>Npj Genomic Medicine</i> , 2019, 4, 27.	3.8	22
23	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. <i>Nature Communications</i> , 2019, 10, 4919.	12.8	111
24	A recurrent missense variant in SLC9A7 causes nonsyndromic X-linked intellectual disability with alteration of Golgi acidification and aberrant glycosylation. <i>Human Molecular Genetics</i> , 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. <i>Translational Psychiatry</i> , 2018, 8, 88.	4.8	22
26	The emerging genetic landscape of cerebral palsy. <i>Handbook of Clinical Neurology</i> / 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. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	6.2	59
28	Pathogenic copy number variants that affect gene expression contribute to genomic burden in cerebral palsy. <i>Npj Genomic Medicine</i> , 2018, 3, 33.	3.8	31
29	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 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. <i>Neurobiology of Disease</i> , 2018, 116, 106-119.	4.4	39
31	Large deletions induced by Cas9 cleavage. <i>Nature</i> , 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 (ER α). <i>Human Molecular Genetics</i> , 2017, 26, 2042-2052.	2.9	28
33	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. <i>European Journal of Medical Genetics</i> , 2017, 60, 437-443.	1.3	10
34	Variant in the X-chromosome spliceosomal gene GPKOW causes male-lethal microcephaly with intrauterine growth restriction. <i>European Journal of Human Genetics</i> , 2017, 25, 1078-1082.	2.8	10
35	A ubiquitin-dependent signalling axis specific for ALKBH-mediated DNA dealkylation repair. <i>Nature</i> , 2017, 551, 389-393.	27.8	83
36	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2. <i>Human Genetics</i> , 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. <i>BMJ Open</i> , 2016, 6, e009537.	1.9	39
38	A non-coding variant in the 5' UTR of <i>DLG3</i> attenuates protein translation to cause non-syndromic intellectual disability. <i>European Journal of Human Genetics</i> , 2016, 24, 1612-1616.	2.8	10
39	Dominant <i>KCNA2</i> mutation causes episodic ataxia and pharmacoresponsive epilepsy. <i>Neurology</i> , 2016, 87, 1975-1984.	1.1	71
40	De Novo Mutations in <i>SLC1A2</i> and <i>CACNA1A</i> Are Important Causes of Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 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. <i>Scientific Reports</i> , 2016, 6, 27010.	3.3	56
42	<i>TBC1D24</i> genotype-phenotype correlation. <i>Neurology</i> , 2016, 87, 77-85.	1.1	97
43	A mutation in <i>COL4A2</i> causes autosomal dominant porencephaly with cataracts. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1059-1063.	1.2	17
44	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. <i>Molecular Psychiatry</i> , 2016, 21, 133-148.	7.9	243
45	Multiplex families with epilepsy. <i>Neurology</i> , 2016, 86, 713-722.	1.1	23
46	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. <i>Human Mutation</i> , 2015, 36, 1197-1204.	2.5	161
47	Increased <i>STAG2</i> dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. <i>Human Molecular Genetics</i> , 2015, 24, 7171-7181.	2.9	28
48	Whole-exome sequencing points to considerable genetic heterogeneity of cerebral palsy. <i>Molecular Psychiatry</i> , 2015, 20, 176-182.	7.9	178
49	A novel X-linked trichothiodystrophy associated with a nonsense mutation in <i>RNF113A</i> . <i>Journal of Medical Genetics</i> , 2015, 52, 269-274.	3.2	302
50	Homozygous mutation of <i>STXBPL</i> explains an autosomal recessive infantile-onset neurodegenerative disorder. <i>Human Molecular Genetics</i> , 2015, 24, 2000-2010.	2.9	25
51	<i>THOC2</i> Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. <i>American Journal of Human Genetics</i> , 2015, 97, 302-310.	6.2	82
52	Developmental disorders: deciphering exomes on a grand scale. <i>Lancet, The</i> , 2015, 385, 1266-1267.	13.7	3
53	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. <i>Human Molecular Genetics</i> , 2015, 24, 5250-5259.	2.9	93
54	Interchromosomal Insertional Translocation at Xq26.3 Alters <i>SOX3</i> Expression in an Individual With XX Male Sex Reversal. <i>Journal of Clinical Endocrinology and Metabolism</i> , 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 F ₂ generation 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 (<i>AUTS2</i>) 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	“North Sea” 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 HUUWE1 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 α -tropomyosin mutation alters dimer preference in nemaline myopathy. Annals of Neurology, 2005, 57, 42-49.	5.3	62
80	A mutation in alpha-tropomyosin slows affects muscle strength, maturation and hypertrophy in a mouse model for nemaline myopathy. Human Molecular Genetics, 2001, 10, 317-328.	2.9	85