Murim Choi

List of Publications by Year
in descending order

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123 papers	24,000 citations	29994 54 h-index	17546 121 g-index
134 all docs	134 docs citations	134 times ranked	39957 citing authors

#	Article	IF	Citations
1	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
2	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. Nature, 2012, 485, 237-241.	13.7	1,863
3	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
4	Genetic diagnosis by whole exome capture and massively parallel DNA sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 19096-19101.	3.3	1,167
5	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
6	Exome sequencing identifies recurrent somatic RAC1 mutations in melanoma. Nature Genetics, 2012, 44, 1006-1014.	9.4	1,052
7	K ⁺ Channel Mutations in Adrenal Aldosterone-Producing Adenomas and Hereditary Hypertension. Science, 2011, 331, 768-772.	6.0	866
8	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	13.7	798
9	Genomic Analysis of Non- NF2 Meningiomas Reveals Mutations in TRAF7 , KLF4 , AKT1 , and SMO. Science, 2013, 339, 1077-1080.	6.0	714
10	Co-occurring Genomic Alterations Define Major Subsets of KRAS-Mutant Lung Adenocarcinoma with Distinct Biology, Immune Profiles, and Therapeutic Vulnerabilities. Cancer Discovery, 2015, 5, 860-877.	7.7	696
11	Mutations in kelch-like 3 and cullin 3 cause hypertension and electrolyte abnormalities. Nature, 2012, 482, 98-102.	13.7	560
12	Genome-wide association study identifies susceptibility loci for IgA nephropathy. Nature Genetics, 2011, 43, 321-327.	9.4	528
13	Somatic and germline CACNA1D calcium channel mutations in aldosterone-producing adenomas and primary aldosteronism. Nature Genetics, 2013, 45, 1050-1054.	9.4	519
14	Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. Nature Genetics, 2014, 46, 1187-1196.	9.4	505
15	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. Nature, 2010, 467, 207-210.	13.7	457
16	Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SeSAME) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 1 the United States of America, 2009, 106, 5842-5847.	3.3	433
17	Recessive mutations in DGKE cause atypical hemolytic-uremic syndrome. Nature Genetics, 2013, 45, 531-536.	9.4	419
18	Mutation of NLRC4 causes a syndrome of enterocolitis and autoinflammation. Nature Genetics, 2014, 46, 1135-1139.	9.4	417

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19	Geographic Differences in Genetic Susceptibility to IgA Nephropathy: GWAS Replication Study and Geospatial Risk Analysis. PLoS Genetics, 2012, 8, e1002765.	1.5	301
20	Characterization of the mutational landscape of anaplastic thyroid cancer via whole-exome sequencing. Human Molecular Genetics, 2015, 24, 2318-2329.	1.4	290
21	Landscape of somatic single-nucleotide and copy-number mutations in uterine serous carcinoma. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 2916-2921.	3.3	275
22	Recurrent gain of function mutation in calcium channel CACNA1H causes early-onset hypertension with primary aldosteronism. ELife, 2015, 4, e06315.	2.8	271
23	Susceptibility loci for intracranial aneurysm in European and Japanese populations. Nature Genetics, 2008, 40, 1472-1477.	9.4	247
24	Morphogenesis of the trachea and esophagus: current players and new roles for noggin and Bmps. Differentiation, 2006, 74, 422-437.	1.0	226
25	Rare copy number variations in congenital heart disease patients identify unique genes in left-right patterning. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 2915-2920.	3.3	226
26	Recurrent activating mutation in PRKACA in cortisol-producing adrenal tumors. Nature Genetics, 2014, 46, 613-617.	9.4	211
27	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. Nature Genetics, 2018, 50, 349-354.	9.4	188
28	Mutational landscape of uterine and ovarian carcinosarcomas implicates histone genes in epithelialâ€“mesenchymal transition. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 12238-12243.	3.3	181
29	Mitotic Recombination in Patients with Ichthyosis Causes Reversion of Dominant Mutations in KRT10. Science, 2010, 330, 94-97.	6.0	176
30	BMP receptor IA is required in mammalian neural crest cells for development of the cardiac outflow tract and ventricular myocardium. Development (Cambridge), 2004, 131, 2205-2218.	1.2	162
31	Whole-exome sequencing and immune profiling of early-stage lung adenocarcinoma with fully annotated clinical follow-up. Annals of Oncology, 2017, 28, 75-82.	0.6	159
32	Comprehensive Re-Sequencing of Adrenal Aldosterone Producing Lesions Reveal Three Somatic Mutations near the KCNJ5 Potassium Channel Selectivity Filter. PLoS ONE, 2012, 7, e41926.	1.1	154
33	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
34	Recessive loss of function of the neuronal ubiquitin hydrolase UCHL1 leads to early-onset progressive neurodegeneration. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 3489-3494.	3.3	144
35	Isolated polycystic liver disease genes define effectors of polycystin-1 function. Journal of Clinical Investigation, 2017, 127, 1772-1785.	3.9	137
36	Identification of Somatic Mutations in Parathyroid Tumors Using Whole-Exome Sequencing. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1774-E1781.	1.8	135

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37	Whole-Exome Sequencing Characterizes the Landscape of Somatic Mutations and Copy Number Alterations in Adrenocortical Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E493-E502.	1.8	131
38	Abatacept alleviates severe autoimmune symptoms in a patient carrying a de novo variant in CTLA-4. Journal of Allergy and Clinical Immunology, 2016, 137, 327-330.	1.5	125
39	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	13.9	119
40	A Form of the Metabolic Syndrome Associated with Mutations inDYRK1B. New England Journal of Medicine, 2014, 370, 1909-1919.	13.9	116
41	Recessive LAMC3 mutations cause malformations of occipital cortical development. Nature Genetics, 2011, 43, 590-594.	9.4	102
42	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	2.6	99
43	Mutation profiles in early-stage lung squamous cell carcinoma with clinical follow-up and correlation with markers of immune function. Annals of Oncology, 2017, 28, 83-89.	0.6	97
44	Taurodontism, variations in tooth number, and misshapened crowns inWnt10anull mice and human kindreds. Molecular Genetics & Genomic Medicine, 2015, 3, 40-58.	0.6	96
45	A logical network-based drug-screening platform for Alzheimerâ€™s disease representing pathological features of human brain organoids. Nature Communications, 2021, 12, 280.	5.8	88
46	Korean Brain Aging Study for the Early Diagnosis and Prediction of Alzheimer's Disease: Methodology and Baseline Sample Characteristics. Psychiatry Investigation, 2017, 14, 851.	0.7	75
47	The Bone Morphogenetic Protein Antagonist Noggin Regulates Mammalian Cardiac Morphogenesis. Circulation Research, 2007, 100, 220-228.	2.0	71
48	Comprehensive DNA methylation analysis of benign and malignant adrenocortical tumors. Genes Chromosomes and Cancer, 2012, 51, 949-960.	1.5	71
49	Tofacitinib relieves symptoms of stimulator of interferon genes (STING)â€“associated vasculopathy with onset in infancy caused by 2 de novo variants in TMEM173. Journal of Allergy and Clinical Immunology, 2017, 139, 1396-1399.e12.	1.5	70
50	Increased Levels of Macrophage Inflammatory Proteins Result in Resistance to R5-Tropic HIV-1 in a Subset of Elite Controllers. Journal of Virology, 2015, 89, 5502-5514.	1.5	68
51	GABBR2 mutations determine phenotype in rett syndrome and epileptic encephalopathy. Annals of Neurology, 2017, 82, 466-478.	2.8	66
52	Endogenous bone morphogenetic protein antagonists regulate mammalian neural crest generation and survival. Developmental Dynamics, 2006, 235, 2507-2520.	0.8	65
53	The BMP antagonist Noggin promotes cranial and spinal neurulation by distinct mechanisms. Developmental Biology, 2006, 295, 647-663.	0.9	63
54	Korean Variant Archive (KOVA): a reference database of genetic variations in the Korean population. Scientific Reports, 2017, 7, 4287.	1.6	60

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55	Neomorphic effects of recurrent somatic mutations in Yin Yang 1 in insulin-producing adenomas. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 4062-4067.	3.3	59
56	Application of Whole Exome Sequencing to Identify Disease-Causing Variants in Inherited Human Diseases. Genomics and Informatics, 2012, 10, 214.	0.4	56
57	ITGB6 loss-of-function mutations cause autosomal recessive amelogenesis imperfecta. Human Molecular Genetics, 2014, 23, 2157-2163.	1.4	54
58	Genetic heterogeneity in Leigh syndrome: Highlighting treatable and novel genetic causes. Clinical Genetics, 2020, 97, 586-594.	1.0	54
59	JAK2, CALR, and MPL mutation spectrum in Japanese patients with myeloproliferative neoplasms. Haematologica, 2015, 100, e46-e48.	1.7	50
60	Geneâ€“environment interactions in severe intraventricular hemorrhage of preterm neonates. Pediatric Research, 2014, 75, 241-250.	1.1	49
61	IL23-Producing Human Lung Cancer Cells Promote Tumor Growth via Conversion of Innate Lymphoid Cell 1 (ILC1) into ILC3. Clinical Cancer Research, 2019, 25, 4026-4037.	3.2	48
62	GM3 synthase deficiency due to ST3GAL5 variants in two Korean female siblings: Masquerading as Rett syndromeâ€like phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2200-2205.	0.7	47
63	Individual exome analysis in diagnosis and management of paediatric liver failure of indeterminate aetiology. Journal of Hepatology, 2014, 61, 1056-1063.	1.8	46
64	Hepatitis B Viral X Protein Overcomes Inhibition of E2F1 Activity by pRb on the HumanRbGene Promoter. DNA and Cell Biology, 2001, 20, 75-80.	0.9	43
65	Phenotype diversity in type 1 Gaucher disease: discovering the genetic basis of Gaucher disease/hematologic malignancy phenotype by individual genome analysis. Blood, 2012, 119, 4731-4740.	0.6	39
66	Taurodeoxycholate Increases the Number of Myeloid-Derived Suppressor Cells That Ameliorate Sepsis in Mice. Frontiers in Immunology, 2018, 9, 1984.	2.2	38
67	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
68	Fam83h null mice support a neomorphic mechanism for human ADHCAI. Molecular Genetics & Genomic Medicine, 2016, 4, 46-67.	0.6	36
69	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. American Journal of Human Genetics, 2017, 101, 824-832.	2.6	36
70	Epigenetic regulation of Kcna3 -encoding Kv1.3 potassium channel by cereblon contributes to regulation of CD4 ⁺ T-cell activation. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 8771-8776.	3.3	35
71	Findings of a 1303 Korean whole-exome sequencing study. Experimental and Molecular Medicine, 2017, 49, e356-e356.	3.2	34
72	Chordin Is a Modifier of Tbx1 for the Craniofacial Malformations of 22q11 Deletion Syndrome Phenotypes in Mouse. PLoS Genetics, 2009, 5, e1000395.	1.5	31

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73	A Genome-Wide Association Study to Identify Single-Nucleotide Polymorphisms for Acute Kidney Injury. American Journal of Respiratory and Critical Care Medicine, 2017, 195, 482-490.	2.5	31
74	Mutations in the Histone Modifier PRDM6 Are Associated with Isolated Nonsyndromic Patent Ductus Arteriosus. American Journal of Human Genetics, 2016, 98, 1082-1091.	2.6	29
75	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
76	Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. Molecular Autism, 2014, 5, 31.	2.6	27
77	Loss of podocalyxin causes a novel syndromic type of congenital nephrotic syndrome. Experimental and Molecular Medicine, 2017, 49, e414-e414.	3.2	27
78	Early-onset generalized dystonia starting in the lower extremities in a patient with a novel ANO3 variant. Parkinsonism and Related Disorders, 2018, 50, 124-125.	1.1	27
79	E2F1 Activates the Human p53 Promoter and Overcomes the Repressive Effect of Hepatitis B Viral X Protein (HBx) on the p53 Promoter. IUBMB Life, 2002, 53, 309-317.	1.5	26
80	Expression patterns of astrocyte elevated gene-1 (AEG-1) during development of the mouse embryo. Gene Expression Patterns, 2010, 10, 361-367.	0.3	26
81	Familial cortical myoclonus with a mutation in NOL3. Annals of Neurology, 2012, 72, 175-183.	2.8	23
82	<scp>LIN</scp> 28A loss of function is associated with Parkinson's disease pathogenesis. EMBO Journal, 2019, 38, e101196.	3.5	23
83	A pilot genome-wide association study shows genomic variants enriched in the non-tumor cells of patients with well-differentiated neuroendocrine tumors of the ileum. Endocrine-Related Cancer, 2011, 18, 171-180.	1.6	22
84	Diagnostic challenge for the rare lysosomal storage disease: Late infantile GM1 gangliosidosis. Brain and Development, 2018, 40, 383-390.	0.6	22
85	Neural-Specific Deletion of Htra2 Causes Cerebellar Neurodegeneration and Defective Processing of Mitochondrial OPA1. PLoS ONE, 2014, 9, e115789.	1.1	21
86	Wiedemann-Steiner Syndrome With 2 Novel KMT2A Mutations. Journal of Child Neurology, 2017, 32, 237-242.	0.7	20
87	Neurogenetic analysis of childhood disintegrative disorder. Molecular Autism, 2017, 8, 19.	2.6	19
88	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
89	Alpha-thalassemia X-linked intellectual disability syndrome identified by whole exome sequencing in two boys with white matter changes and developmental retardation. Gene, 2015, 569, 318-322.	1.0	18
90	The dentin phosphoprotein repeat region and inherited defects of dentin. Molecular Genetics & Genomic Medicine, 2016, 4, 28-38.	0.6	18

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91	Kidney residency of VISTA-positive macrophages accelerates repair from ischemic injury. Kidney International, 2020, 97, 980-994.	2.6	18
92	On optimal pooling designs to identify rare variants through massive resequencing. Genetic Epidemiology, 2011, 35, 139-147.	0.6	16
93	Rare cases of congenital arthrogryposis multiplex caused by novel recurrent CHRNG mutations. Journal of Human Genetics, 2015, 60, 213-215.	1.1	16
94	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. American Journal of Human Genetics, 2019, 104, 439-453.	2.6	16
95	Disease-specific eQTL screening reveals an anti-fibrotic effect of AGXT2 in non-alcoholic fatty liver disease. Journal of Hepatology, 2021, 75, 514-523.	1.8	16
96	Diagnostic Challenges Associated with GLUT1 Deficiency: Phenotypic Variabilities and Evolving Clinical Features. Yonsei Medical Journal, 2019, 60, 1209.	0.9	16
97	Atypical presentation of infantileâ€onset farber disease with novel ASAH1 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3023-3027.	0.7	15
98	Root anomalies and dentin dysplasia in autosomal recessive hyperphosphatemic familial tumoral calcinosis (HFTC). Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2015, 120, e235-e239.	0.2	13
99	Somatic uniparental disomy mitigates the most damaging EFL1 allele combination in Shwachman-Diamond syndrome. Blood, 2021, 138, 2117-2128.	0.6	13
100	SOFT syndrome caused by compound heterozygous mutations of POC1A and its skeletal manifestation. Journal of Human Genetics, 2016, 61, 561-564.	1.1	11
101	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	2.6	11
102	Genetic associations of in vivo pathology influence Alzheimerâ€™s disease susceptibility. Alzheimer's Research and Therapy, 2020, 12, 156.	3.0	11
103	Generation and characterization of a mitotane-resistant adrenocortical cell line. Endocrine Connections, 2020, 9, 122-134.	0.8	11
104	Frequency data on four tetrameric STR loci D18S1270, D14S608, D16S3253 and D21S1437 in a Korean population. International Journal of Legal Medicine, 2000, 113, 179-180.	1.2	10
105	Familial cases of progressive myoclonic epilepsy caused by maternal somatic mosaicism of a recurrent KCNC1 p.Arg320His mutation. Brain and Development, 2018, 40, 429-432.	0.6	10
106	Multiâ€Omicsâ€Based Autophagyâ€Related Untypical Subtypes in Patients with Cerebral Amyloid Pathology. Advanced Science, 2022, 9, .	5.6	9
107	Oncogenic effects of germline variants in lysosomal storage disease genes. Genetics in Medicine, 2019, 21, 2695-2705.	1.1	7
108	Overexpression of Replication-Dependent Histone Signifies a Subset of Dedifferentiated Liposarcoma with Increased Aggressiveness. Cancers, 2021, 13, 3122.	1.7	7

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109	Ultra-rare Disease and Genomics-Driven Precision Medicine. Genomics and Informatics, 2016, 14, 42.	0.4	6
110	Transcriptome-based variant calling and aberrant mRNA discovery enhance diagnostic efficiency for neuromuscular diseases. Journal of Medical Genetics, 2022, 59, 1075-1081.	1.5	6
111	eIF2B-related multisystem disorder in two sisters with atypical presentations. European Journal of Paediatric Neurology, 2017, 21, 404-409.	0.7	4
112	Biallelic mutations in ABCB1 display recurrent reversible encephalopathy. Annals of Clinical and Translational Neurology, 2020, 7, 1443-1449.	1.7	4
113	A Familial Case of Childhood Ataxia with Leukodystrophy Due to Novel POLR1C Mutations.		