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	Impact of Evolutionary Changes in Nonalcoholic Fatty Liver Disease on Lung Function Decline. Gut and Liver, 2023, 17, 139-149.	1.4	1
2	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
3	Multiâ€Omicsâ€Based Autophagyâ€Related Untypical Subtypes in Patients with Cerebral Amyloid Pathology. Advanced Science, 2022, 9, .	5.6	9
4	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
5	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
6	Somatic uniparental disomy mitigates the most damaging EFL1 allele combination in Shwachman-Diamond syndrome. Blood, 2021, 138, 2117-2128.	0.6	13
7	Overexpression of Replication-Dependent Histone Signifies a Subset of Dedifferentiated Liposarcoma with Increased Aggressiveness. Cancers, 2021, 13, 3122.	1.7	7
8	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
9	A networkâ€based drugâ€screening platform for Alzheimerâ€™s disease by integrating mathematical modeling and pathological features of human brain organoids. Alzheimer's and Dementia, 2021, 17, .	0.4	0
10	Kidney residency of VISTA-positive macrophages accelerates repair from ischemic injury. Kidney International, 2020, 97, 980-994.	2.6	18
11	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
12	Genetic associations of in vivo pathology influence Alzheimerâ€™s disease susceptibility. Alzheimer's Research and Therapy, 2020, 12, 156.	3.0	11
13	Identifying germline APOBEC3B deletion and immune phenotype in Korean patients with operable breast cancer. Breast Cancer Research and Treatment, 2020, 183, 697-704.	1.1	3
14	Biallelic mutations in ABCB1 display recurrent reversible encephalopathy. Annals of Clinical and Translational Neurology, 2020, 7, 1443-1449.	1.7	4
15	Genetic heterogeneity in Leigh syndrome: Highlighting treatable and novel genetic causes. Clinical Genetics, 2020, 97, 586-594.	1.0	54
16	Generation and characterization of a mitotane-resistant adrenocortical cell line. Endocrine Connections, 2020, 9, 122-134.	0.8	11
17	A Familial Case of Childhood Ataxia with Leukodystrophy Due to Novel POLR1C Mutations.		

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19	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
20	Oncogenic effects of germline variants in lysosomal storage disease genes. Genetics in Medicine, 2019, 21, 2695-2705.	1.1	7
21	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
22	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
23	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
24	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. American Journal of Human Genetics, 2019, 104, 439-453.	2.6	16
25	<scp>LIN</scp> 28A loss of function is associated with Parkinson's disease pathogenesis. EMBO Journal, 2019, 38, e101196.	3.5	23
26	Diagnostic Challenges Associated with GLUT1 Deficiency: Phenotypic Variabilities and Evolving Clinical Features. Yonsei Medical Journal, 2019, 60, 1209.	0.9	16
27	Clinical and genetic characteristics of Korean patients with IARS2-related disorders. Journal of Genetic Medicine, 2019, 16, 55-61.	0.1	4
28	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
29	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
30	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. Nature Genetics, 2018, 50, 349-354.	9.4	188
31	Genomic analysis of synchronous intracranial meningiomas with different histological grades. Journal of Neuro-Oncology, 2018, 138, 41-48.	1.4	2
32	Diagnostic challenge for the rare lysosomal storage disease: Late infantile GM1 gangliosidosis. Brain and Development, 2018, 40, 383-390.	0.6	22
33	Reply to â€œa novel mutation in the transmembrane 6 domain of GABBR2 leads to a rettâ€like phenotypeâ€. Annals of Neurology, 2018, 83, 439-439.	2.8	1
34	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	2.6	99
35	Taurodeoxycholate Increases the Number of Myeloid-Derived Suppressor Cells That Ameliorate Sepsis in Mice. Frontiers in Immunology, 2018, 9, 1984.	2.2	38
36	A family with NKX2.5 gene mutations presenting as familial atrial septal defect and atrioventricular block: A case report. Journal of Genetic Medicine, 2018, 15, 20-23.	0.1	0

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37	Neurogenetic analysis of childhood disintegrative disorder. Molecular Autism, 2017, 8, 19.	2.6	19
38	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
39	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
40	GABBR2 mutations determine phenotype in rett syndrome and epileptic encephalopathy. Annals of Neurology, 2017, 82, 466-478.	2.8	66
41	Findings of a 1303 Korean whole-exome sequencing study. Experimental and Molecular Medicine, 2017, 49, e356-e356.	3.2	34
42	Loss of podocalyxin causes a novel syndromic type of congenital nephrotic syndrome. Experimental and Molecular Medicine, 2017, 49, e414-e414.	3.2	27
43	Severe hypotonia and postnatal growth impairment in a girl with a missense mutation in COL1A1 : Implication of expanded phenotypic spectrum of type I collagenopathy. Brain and Development, 2017, 39, 799-803.	0.6	3
44	Korean Variant Archive (KOVA): a reference database of genetic variations in the Korean population. Scientific Reports, 2017, 7, 4287.	1.6	60
45	Wiedemann-Steiner Syndrome With 2 Novel KMT2A Mutations. Journal of Child Neurology, 2017, 32, 237-242.	0.7	20
46	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
47	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
48	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
49	eIF2B-related multisystem disorder in two sisters with atypical presentations. European Journal of Paediatric Neurology, 2017, 21, 404-409.	0.7	4
50	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
51	Isolated polycystic liver disease genes define effectors of polycystin-1 function. Journal of Clinical Investigation, 2017, 127, 1772-1785.	3.9	137
52	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
53	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
54	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

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55	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
56	The dentin phosphoprotein repeat region and inherited defects of dentin. Molecular Genetics & Genomic Medicine, 2016, 4, 28-38.	0.6	18
57	Fam83h null mice support a neomorphic mechanism for human ADHCAI. Molecular Genetics & Genomic Medicine, 2016, 4, 46-67.	0.6	36
58	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
59	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
60	SOFT syndrome caused by compound heterozygous mutations of POC1A and its skeletal manifestation. Journal of Human Genetics, 2016, 61, 561-564.	1.1	11
61	Ultra-rare Disease and Genomics-Driven Precision Medicine. Genomics and Informatics, 2016, 14, 42.	0.4	6
62	Taurodontism, variations in tooth number, and misshapened crowns inWnt10anull mice and human kindreds. Molecular Genetics & Genomic Medicine, 2015, 3, 40-58.	0.6	96
63	Recurrent gain of function mutation in calcium channel CACNA1H causes early-onset hypertension with primary aldosteronism. ELife, 2015, 4, e06315.	2.8	271
64	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
65	Rare cases of congenital arthrogryposis multiplex caused by novel recurrent CHRNG mutations. Journal of Human Genetics, 2015, 60, 213-215.	1.1	16
66	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
67	JAK2, CALR, and MPL mutation spectrum in Japanese patients with myeloproliferative neoplasms. Haematologica, 2015, 100, e46-e48.	1.7	50
68	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
69	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
70	Characterization of the mutational landscape of anaplastic thyroid cancer via whole-exome sequencing. Human Molecular Genetics, 2015, 24, 2318-2329.	1.4	290
71	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
72	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

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73	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
74	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
75	The role of de novo variants in complex and rare diseases pathogenesis. Journal of Genetic Medicine, 2015, 12, 1-5.	0.1	1
76	Neural-Specific Deletion of Htra2 Causes Cerebellar Neurodegeneration and Defective Processing of Mitochondrial OPA1. PLoS ONE, 2014, 9, e115789.	1.1	21
77	Geneâ€“environment interactions in severe intraventricular hemorrhage of preterm neonates. Pediatric Research, 2014, 75, 241-250.	1.1	49
78	Recurrent activating mutation in PRKACA in cortisol-producing adrenal tumors. Nature Genetics, 2014, 46, 613-617.	9.4	211
79	A Form of the Metabolic Syndrome Associated with Mutations inDYRK1B. New England Journal of Medicine, 2014, 370, 1909-1919.	13.9	116
80	ITGB6 loss-of-function mutations cause autosomal recessive amelogenesis imperfecta. Human Molecular Genetics, 2014, 23, 2157-2163.	1.4	54
81	Mutation of NLRC4 causes a syndrome of enterocolitis and autoinflammation. Nature Genetics, 2014, 46, 1135-1139.	9.4	417
82	Individual exome analysis in diagnosis and management of paediatric liver failure of indeterminate aetiology. Journal of Hepatology, 2014, 61, 1056-1063.	1.8	46
83	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
84	Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. Molecular Autism, 2014, 5, 31.	2.6	27
85	Somatic and germline CACNA1D calcium channel mutations in aldosterone-producing adenomas and primary aldosteronism. Nature Genetics, 2013, 45, 1050-1054.	9.4	519
86	Genomic Analysis of Non- NF2 Meningiomas Reveals Mutations in TRAF7 , KLF4 , AKT1 , and SMO. Science, 2013, 339, 1077-1080.	6.0	714
87	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
88	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
89	Recessive mutations in DGKE cause atypical hemolytic-uremic syndrome. Nature Genetics, 2013, 45, 531-536.	9.4	419
90	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	13.7	798

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91	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	13.9	119
92	Geographic Differences in Genetic Susceptibility to IgA Nephropathy: GWAS Replication Study and Geospatial Risk Analysis. PLoS Genetics, 2012, 8, e1002765.	1.5	301
93	Mutations in kelch-like 3 and cullin 3 cause hypertension and electrolyte abnormalities. Nature, 2012, 482, 98-102.	13.7	560
94	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
95	Identification of Somatic Mutations in Parathyroid Tumors Using Whole-Exome Sequencing. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1774-E1781.	1.8	135
96	Exome sequencing identifies recurrent somatic RAC1 mutations in melanoma. Nature Genetics, 2012, 44, 1006-1014.	9.4	1,052
97	Familial cortical myoclonus with a mutation in NOL3. Annals of Neurology, 2012, 72, 175-183.	2.8	23
98	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. Nature, 2012, 485, 237-241.	13.7	1,863
99	Comprehensive DNA methylation analysis of benign and malignant adrenocortical tumors. Genes Chromosomes and Cancer, 2012, 51, 949-960.	1.5	71
100	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
101	Application of Whole Exome Sequencing to Identify Disease-Causing Variants in Inherited Human Diseases. Genomics and Informatics, 2012, 10, 214.	0.4	56
102	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
103	Genome-wide association study identifies susceptibility loci for IgA nephropathy. Nature Genetics, 2011, 43, 321-327.	9.4	528
104	Recessive LAMC3 mutations cause malformations of occipital cortical development. Nature Genetics, 2011, 43, 590-594.	9.4	102
105	On optimal pooling designs to identify rare variants through massive resequencing. Genetic Epidemiology, 2011, 35, 139-147.	0.6	16
106	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
107	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
108	K ⁺ Channel Mutations in Adrenal Aldosterone-Producing Adenomas and Hereditary Hypertension. Science, 2011, 331, 768-772.	6.0	866

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109	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
110	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. Nature, 2010, 467, 207-210.	13.7	457
111	Mitotic Recombination in Patients with Ichthyosis Causes Reversion of Dominant Mutations in KRT10. Science, 2010, 330, 94-97.	6.0	176
112	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
113	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
114	Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SeSAME) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 5 the United States of America, 2009, 106, 5842-5847.	3.3	433
115	Susceptibility loci for intracranial aneurysm in European and Japanese populations. Nature Genetics, 2008, 40, 1472-1477.	9.4	247
116	The Bone Morphogenetic Protein Antagonist Noggin Regulates Mammalian Cardiac Morphogenesis. Circulation Research, 2007, 100, 220-228.	2.0	71
117	The BMP antagonist Noggin promotes cranial and spinal neurulation by distinct mechanisms. Developmental Biology, 2006, 295, 647-663.	0.9	63
118	Morphogenesis of the trachea and esophagus: current players and new roles for noggin and Bmps. Differentiation, 2006, 74, 422-437.	1.0	226
119	Endogenous bone morphogenetic protein antagonists regulate mammalian neural crest generation and survival. Developmental Dynamics, 2006, 235, 2507-2520.	0.8	65
120	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
121	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
122	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
123	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