Wendy Chung

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8826911/publications.pdf

Version: 2024-02-01

		2802	3261
586	46,279	94	185
papers	citations	h-index	g-index

621	621	621	54407
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Strong Association of De Novo Copy Number Mutations with Autism. Science, 2007, 316, 445-449.	12.6	2,497
2	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. JAMA - Journal of the American Medical Association, 2017, 317, 2402.	7.4	1,898
3	Induced Pluripotent Stem Cells Generated from Patients with ALS Can Be Differentiated into Motor Neurons. Science, 2008, 321, 1218-1221.	12.6	1,826
4	Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2017, 19, 249-255.	2.4	1,398
5	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	21.4	1,141
6	Pediatric Pulmonary Hypertension. Circulation, 2015, 132, 2037-2099.	1.6	879
7	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
8	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	27.8	798
9	Clinical application of whole-exome sequencing across clinical indications. Genetics in Medicine, 2016, 18, 696-704.	2.4	780
10	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	12.6	646
11	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	21.4	624
12	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115.	0.6	584
13	A Novel Channelopathy in Pulmonary Arterial Hypertension. New England Journal of Medicine, 2013, 369, 351-361.	27.0	412
14	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
15	Genetic Basis for Congenital Heart Disease: Revisited: A Scientific Statement From the American Heart Association. Circulation, 2018, 138, e653-e711.	1.6	387
16	Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 2413-2421.	2.4	378
17	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756.	27.8	369
18	Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2013, 62, D13-D21.	2.8	367

#	Article	IF	Citations
19	Observational study of spinal muscular atrophy type I and implications for clinical trials. Neurology, 2014, 83, 810-817.	1.1	367
20	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
21	ACMG SF v3.0 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1381-1390.	2.4	356
22	Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2009, 54, S32-S42.	2.8	342
23	Return of Genomic Results to Research Participants: The Floor, the Ceiling, and the Choices In Between. American Journal of Human Genetics, 2014, 94, 818-826.	6.2	342
24	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
25	Whole Exome Sequencing to Identify a Novel Gene (Caveolin-1) Associated With Human Pulmonary Arterial Hypertension. Circulation: Cardiovascular Genetics, 2012, 5, 336-343.	5.1	333
26	Glucocerebrosidase activity in Parkinsonâ€™s disease with and withoutGBAmutations. Brain, 2015, 138, 2648-2658.	7.6	326
27	Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2014, 16, e1-e29.	2.4	318
28	Analysis of GNAS mutations in 60 growth hormone secreting pituitary tumors: correlation with clinical and pathological characteristics and surgical outcome based on highly sensitive GH and IGF-I criteria for remission. Pituitary, 2007, 10, 275-282.	2.9	316
29	BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. Lancet Respiratory Medicine,the, 2016, 4, 129-137.	10.7	307
30	Genetics and genomics of pulmonary arterial hypertension. European Respiratory Journal, 2019, 53, 1801899.	6.7	306
31	MC4R-dependent suppression of appetite by bone-derived lipocalin 2. Nature, 2017, 543, 385-390.	27.8	299
32	Enalapril in Infants With Single Ventricle. Circulation, 2010, 122, 333-340.	1.6	267
33	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493.	8.1	265
34	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
35	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
36	A 600â€…kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. Journal of Medical Genetics, 2012, 49, 660-668.	3.2	251

#	Article	IF	Citations
37	The Molecular Genetics of Rodent Single Gene Obesities. Journal of Biological Chemistry, 1997, 272, 31937-31940.	3.4	245
38	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
39	Glycogen Storage Disease Type III diagnosis and management guidelines. Genetics in Medicine, 2010, 12, 446-463.	2.4	236
40	Efficacy and safety of setmelanotide, an MC4R agonist, in individuals with severe obesity due to LEPR or POMC deficiency: single-arm, open-label, multicentre, phase 3 trials. Lancet Diabetes and Endocrinology,the, 2020, 8, 960-970.	11.4	235
41	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
42	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. Circulation Research, 2014, 115, 884-896.	4.5	229
43	Pathogenic and likely pathogenic variant prevalence among the first 10,000 patients referred for next-generation cancer panel testing. Genetics in Medicine, 2016, 18, 823-832.	2.4	227
44	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
45	RASA1Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. Human Mutation, 2013, 34, 1632-1641.	2.5	221
46	Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456.	8.6	210
47	Pediatric Cardiomyopathies. Circulation Research, 2017, 121, 855-873.	4.5	207
48	Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling. Circulation, 2017, 136, 1037-1048.	1.6	204
49	Precision Medicine in Diabetes: A Consensus Report From the American Diabetes Association (ADA) and the European Association for the Study of Diabetes (EASD). Diabetes Care, 2020, 43, 1617-1635.	8.6	204
50	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	6.2	201
51	The Cognitive and Behavioral Phenotype of the 16p11.2 Deletion in a Clinically Ascertained Population. Biological Psychiatry, 2015, 77, 785-793.	1.3	198
52	The usefulness of whole-exome sequencing in routine clinical practice. Genetics in Medicine, 2014, 16, 922-931.	2.4	196
53	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
54	A public resource facilitating clinical use of genomes. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 11920-11927.	7.1	194

#	Article	IF	Citations
55	Pancreatic Cancer Screening in a Prospective Cohort of High-Risk Patients: A Comprehensive Strategy of Imaging and Genetics. Clinical Cancer Research, 2010, 16, 5028-5037.	7.0	189
56	Induced pluripotent stem cells used to reveal drug actions in a long QT syndrome family with complex genetics. Journal of General Physiology, 2013, 141, 61-72.	1.9	189
57	Fatal infantile neuromuscular presentation of glycogen storage disease type IV. Neuromuscular Disorders, 2004, 14, 253-260.	0.6	185
58	Outcomes of Congenital Diaphragmatic Hernia in the Modern Era ofÂManagement. Journal of Pediatrics, 2013, 163, 114-119.e1.	1.8	185
59	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	2.5	185
60	EIF2AK4 Mutations in Pulmonary Capillary Hemangiomatosis. Chest, 2014, 145, 231-236.	0.8	176
61	Comparison of Parkinson Risk in Ashkenazi Jewish Patients With Gaucher Disease andGBAHeterozygotes. JAMA Neurology, 2014, 71, 752.	9.0	172
62	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	3.8	163
63	Opposing Brain Differences in 16p11.2 Deletion and Duplication Carriers. Journal of Neuroscience, 2014, 34, 11199-11211.	3.6	149
64	Attitudes and Practices Among Internists Concerning Genetic Testing. Journal of Genetic Counseling, 2013, 22, 90-100.	1.6	147
65	Implementation of next generation sequencing into pediatric hematology-oncology practice: moving beyond actionable alterations. Genome Medicine, 2016, 8, 133.	8.2	147
66	Points to consider in the reevaluation and reanalysis of genomic test results: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 1267-1270.	2.4	147
67	Genetic Testing for Dilated Cardiomyopathy in Clinical Practice. Journal of Cardiac Failure, 2012, 18, 296-303.	1.7	145
68	Hypomorphism for RPGRIP1L, a Ciliary Gene Vicinal to the FTO Locus, Causes Increased Adiposity in Mice. Cell Metabolism, 2014, 19, 767-779.	16.2	145
69	Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2021 update: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1391-1398.	2.4	145
70	Î²-Cell Dysfunction Due to Increased ER Stress in a Stem Cell Model of Wolfram Syndrome. Diabetes, 2014, 63, 923-933.	0.6	144
71	Validation of the Expanded Hammersmith Functional Motor Scale in Spinal Muscular Atrophy Type II and III. Journal of Child Neurology, 2011, 26, 1499-1507.	1.4	143
72	Congenital diaphragmatic hernias: from genes to mechanisms to therapies. DMM Disease Models and Mechanisms, 2017, 10, 955-970.	2.4	143

#	Article	IF	Citations
73	Observational Study of Spinal Muscular Atrophy Type 2 and 3. Archives of Neurology, 2011, 68, 779.	4.5	142
74	The Congenital Heart Disease Genetic Network Study. Circulation Research, 2013, 112, 698-706.	4.5	142
75	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
76	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
77	A Recurrent PDGFRB Mutation Causes Familial Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 996-1000.	6.2	135
78	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	6.2	134
79	Clinical phenotype of the recurrent 1q21.1 copy-number variant. Genetics in Medicine, 2016, 18, 341-349.	2.4	134
80	Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. Journal of Clinical Oncology, 2016, 34, 2172-2181.	1.6	132
81	16p11.2 deletion and duplication: Characterizing neurologic phenotypes in a large clinically ascertained cohort. American Journal of Medical Genetics, Part A, 2016, 170, 2943-2955.	1.2	131
82	Fine Structure of the Murine Leptin Receptor Gene: Splice Site Suppression Is Required to Form Two Alternatively Spliced Transcripts. Genomics, 1997, 45, 264-270.	2.9	130
83	Cut-like Homeobox 1 (CUX1) Regulates Expression of the Fat Mass and Obesity-associated and Retinitis Pigmentosa GTPase Regulator-interacting Protein-1-like (RPGRIP1L) Genes and Coordinates Leptin Receptor Signaling. Journal of Biological Chemistry, 2011, 286, 2155-2170.	3.4	129
84	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444.	6.2	124
85	CANOES: detecting rare copy number variants from whole exome sequencing data. Nucleic Acids Research, 2014, 42, e97-e97.	14.5	123
86	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
87	ACMG SF v3.1 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 1407-1414.	2.4	119
88	Complex Genetics and the Etiology of Human Congenital Heart Disease. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a013953-a013953.	6.2	118
89	MSH6 and PMS2 germ-line pathogenic variants implicated in Lynch syndrome are associated with breast cancer. Genetics in Medicine, 2018, 20, 1167-1174.	2.4	116
90	10-year performance of four models of breast cancer risk: a validation study. Lancet Oncology, The, 2019, 20, 504-517.	10.7	116

#	Article	IF	Citations
91	PVDOMICS. Circulation Research, 2017, 121, 1136-1139.	4.5	113
92	The contribution of de novo and rare inherited copy number changes to congenital heart disease in an unselected sample of children with conotruncal defects or hypoplastic left heart disease. Human Genetics, 2014, 133, 11-27.	3.8	112
93	The Role of Parental Cognitive, Behavioral, and Motor Profiles in Clinical Variability in Individuals With Chromosome 16p11.2 Deletions. JAMA Psychiatry, 2015, 72, 119.	11.0	112
94	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	8.2	112
95	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	6.2	110
96	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
97	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
98	Validation of the Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	0.6	106
99	Rare variant phasing and haplotypic expression from RNA sequencing with phASER. Nature Communications, 2016, 7, 12817.	12.8	105
100	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
101	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	3.6	104
102	Researchersâ€™ views on return of incidental genomic research results: qualitative and quantitative findings. Genetics in Medicine, 2013, 15, 888-895.	2.4	103
103	Genetic loss of SH2B3 in acute lymphoblastic leukemia. Blood, 2013, 122, 2425-2432.	1.4	101
104	Deep Phenotyping on Electronic Health Records Facilitates Genetic Diagnosis by Clinical Exomes. American Journal of Human Genetics, 2018, 103, 58-73.	6.2	99
105	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605.	6.2	99
106	Pilot study of population-based newborn screening for spinal muscular atrophy in New York state. Genetics in Medicine, 2018, 20, 608-613.	2.4	98
107	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	21.4	97
108	Brief Report: Impact of COVID-19 on Individuals with ASD and Their Caregivers: A Perspective from the SPARK Cohort. Journal of Autism and Developmental Disorders, 2021, 51, 3766-3773.	2.7	97

#	Article	IF	Citations
109	Genome-wide association analysis identifies a susceptibility locus for pulmonary arterial hypertension. Nature Genetics, 2013, 45, 518-521.	21.4	93
110	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
111	Psychiatric disorders in children with 16p11.2 deletion and duplication. Translational Psychiatry, 2019, 9, 8.	4.8	93
112	Serum Endostatin Is a Genetically Determined Predictor of Survival in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2015, 191, 208-218.	5.6	92
113	Impact of Panel Gene Testing for Hereditary Breast and Ovarian Cancer on Patients. Journal of Genetic Counseling, 2017, 26, 1116-1129.	1.6	90
114	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
115	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
116	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
117	Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. Circulation: Cardiovascular Genetics, 2013, 6, 444-451.	5.1	89
118	Congenital Disorder of Glycosylation Id Presenting with Hyperinsulinemic Hypoglycemia and Islet Cell Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 4371-4375.	3.6	88
119	Use of Genetic Tests among Neurologists and Psychiatrists: Knowledge, Attitudes, Behaviors, and Needs for Training. Journal of Genetic Counseling, 2014, 23, 156-163.	1.6	87
120	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69.	8.2	86
121	MVP predicts theÂpathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	12.8	85
122	iPSC-derived Î² cells model diabetes due to glucokinase deficiency. Journal of Clinical Investigation, 2013, 123, 3146-3153.	8.2	84
123	Glut1 deficiency syndrome and erythrocyte glucose uptake assay. Annals of Neurology, 2011, 70, 996-1005.	5.3	83
124	Risk of Pancreatic Cancer in Breast Cancer Families from the Breast Cancer Family Registry. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 803-811.	2.5	83
125	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
126	Advances in the Understanding of the Genetic Determinants of Congenital Heart Disease and Their Impact on Clinical Outcomes. Journal of the American Heart Association, 2018, 7, .	3.7	82

#	Article	IF	Citations
127	The Congenital Heart Disease Genetic Network Study: Cohort description. PLoS ONE, 2018, 13, e0191319.	2.5	82
128	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	2.4	82
129	Variants in GATA4 are a rare cause of familial and sporadic congenital diaphragmatic hernia. Human Genetics, 2013, 132, 285-292.	3.8	81
130	SMPD1 mutations, activity, and Î±â€synuclein accumulation in Parkinson's disease. Movement Disorders, 2019, 34, 526-535.	3.9	81
131	Diagnosis and management of glycogen storage diseases type VI and IX: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 772-789.	2.4	81
132	Spectrum of Neuropathophysiology in Spinal Muscular Atrophy Type I. Journal of Neuropathology and Experimental Neurology, 2015, 74, 15-24.	1.7	80
133	Development of a tiered and binned genetic counseling model for informed consent in the era of multiplex testing for cancer susceptibility. Genetics in Medicine, 2015, 17, 485-492.	2.4	79
134	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. PLoS Genetics, 2018, 14, e1007822.	3.5	79
135	Glut1 deficiency: Inheritance pattern determined by haploinsufficiency. Annals of Neurology, 2010, 68, 955-958.	5.3	78
136	Genetic causes of congenital diaphragmatic hernia. Seminars in Fetal and Neonatal Medicine, 2014, 19, 324-330.	2.3	77
137	Genetic evaluation and counseling for epilepsy. Nature Reviews Neurology, 2010, 6, 445-453.	10.1	75
138	The genetic basis of pulmonary arterial hypertension. Human Genetics, 2014, 133, 471-479.	3.8	75
139	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	6.2	75
140	Novel Mutations and Decreased Expression of the Epigenetic Regulator TET2 in Pulmonary Arterial Hypertension. Circulation, 2020, 141, 1986-2000.	1.6	75
141	Amino Acid Variants in the Human Leptin Receptor: Lack of Association to Juvenile Onset Obesity. Biochemical and Biophysical Research Communications, 1997, 233, 248-252.	2.1	74
142	Copy number variants and infantile spasms: evidence for abnormalities in ventral forebrain development and pathways of synaptic function. European Journal of Human Genetics, 2011, 19, 1238-1245.	2.8	74
143	CSER and eMERGE: current and potential state of the display of genetic information in the electronic health record. Journal of the American Medical Informatics Association: JAMIA, 2015, 22, 1231-1242.	4.4	73
144	Aberrant White Matter Microstructure in Children with 16p11.2 Deletions. Journal of Neuroscience, 2014, 34, 6214-6223.	3.6	70

#	Article	IF	Citations
145	A complete deficiency of Hyaluronoglucosaminidase 1 (HYAL1) presenting as familial juvenile idiopathic arthritis. Journal of Inherited Metabolic Disease, 2011, 34, 1013-1022.	3.6	68
146	De novo copy number variants are associated with congenital diaphragmatic hernia. Journal of Medical Genetics, 2012, 49, 650-659.	3.2	68
147	Return of Secondary Genomic Findings vs Patient Autonomy. JAMA - Journal of the American Medical Association, 2013, 310, 369.	7.4	68
148	Developmental outcomes of children with congenital diaphragmatic hernia: A multicenter prospective study. Journal of Pediatric Surgery, 2013, 48, 1995-2004.	1.6	68
149	Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547.	3.2	68
150	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	2.4	68
151	Absence epilepsy in apathetic, a spontaneous mutant mouse lacking the h channel subunit, HCN2. Neurobiology of Disease, 2009, 33, 499-508.	4.4	67
152	Short Communication: The Cardiac Myosin Binding Protein C Arg502Trp Mutation. Circulation Research, 2010, 106, 1549-1552.	4.5	67
153	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	6.2	66
154	High Prevalence of BRCA1 and BRCA2 Germline Mutations with Loss of Heterozygosity in a Series of Resected Pancreatic Adenocarcinoma and Other Neoplastic Lesions. Clinical Cancer Research, 2013, 19, 3396-3403.	7.0	65
155	Increased burden ofde novopredicted deleterious variants in complex congenital diaphragmatic hernia. Human Molecular Genetics, 2015, 24, 4764-4773.	2.9	65
156	Brief Report: SETD2 Mutation in a Child with Autism, Intellectual Disabilities and Epilepsy. Journal of Autism and Developmental Disorders, 2015, 45, 3764-3770.	2.7	64
157	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	2.4	64
158	A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. Nature Communications, 2018, 9, 67.	12.8	64
159	Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1399-1415.	2.4	64
160	Molecular and functional analysis ofSLC25A20 mutations causing carnitine-acylcarnitine translocase deficiency. Human Mutation, 2004, 24, 312-320.	2.5	63
161	Decision-Making About Reproductive Choices Among Individuals At-Risk for Huntington's Disease. Journal of Genetic Counseling, 2007, 16, 347-362.	1.6	63
162	Renin-Angiotensin-Aldosterone Genotype Influences Ventricular Remodeling in Infants With Single Ventricle. Circulation, 2011, 123, 2353-2362.	1.6	63

#	Article	IF	Citations
163	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
164	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	6.2	62
165	Loss-of-Function ABCC8 Mutations in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002087.	3.6	62
166	A Genetics-First Approach to Dissecting the Heterogeneity of Autism: Phenotypic Comparison of Autism Risk Copy Number Variants. American Journal of Psychiatry, 2021, 178, 77-86.	7.2	62
167	De novo missense variants in PPP2R5D are associated with intellectual disability, macrocephaly, hypotonia, and autism. Neurogenetics, 2016, 17, 43-49.	1.4	61
168	Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477.	2.4	61
169	FTO genotype impacts food intake and corticolimbic activation. American Journal of Clinical Nutrition, 2018, 107, 145-154.	4.7	60
170	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	3.9	60
171	Defining a comprehensive verotype using electronic health records for personalized medicine. Journal of the American Medical Informatics Association: JAMIA, 2013, 20, e232-e238.	4.4	59
172	BRCA1 and BRCA2 germline mutations are frequently demonstrated in both highâ€risk pancreatic cancer screening and pancreatic cancer cohorts. Cancer, 2014, 120, 1960-1967.	4.1	59
173	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
174	Positional Cloning of â€œLisch-likeâ€, a Candidate Modifier of Susceptibility to Type 2 Diabetes in Mice. PLoS Genetics, 2008, 4, e1000137.	3.5	58
175	Informed consent for return of incidental findings in genomic research. Genetics in Medicine, 2014, 16, 367-373.	2.4	58
176	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. Nature Communications, 2019, 10, 4722.	12.8	58
177	Genetic Modifiers ofLeprfaAssociated with Variability in Insulin Production and Susceptibility to NIDDM. Genomics, 1997, 41, 332-344.	2.9	57
178	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	1.2	57
179	Implementation of population-based newborn screening reveals low incidence of spinal muscular atrophy. Genetics in Medicine, 2020, 22, 1296-1302.	2.4	57
180	Suggestive Linkages Between Markers on Human 1p32â€p22 and Body Fat and Insulin Levels in the QuÃ©bec Family Study. Obesity, 1997, 5, 115-121.	4.0	56

#	Article	IF	Citations
181	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	1.3	56
182	Mutation inSNAP25as a novel genetic cause of epilepsy and intellectual disability. Rare Diseases (Austin, Tex), 2013, 1, e26314.	1.8	55
183	Whole exome sequencing identifies de novo mutations inGATA6associated with congenital diaphragmatic hernia. Journal of Medical Genetics, 2014, 51, 197-202.	3.2	55
184	Psychological outcomes related to exome and genome sequencing result disclosure: a meta-analysis of seven Clinical Sequencing Exploratory Research (CSER) Consortium studies. Genetics in Medicine, 2019, 21, 2781-2790.	2.4	55
185	Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314.	1.4	54
186	Association of Plastin 3 Expression With Disease Severity in Spinal Muscular Atrophy Only in Postpubertal Females. Archives of Neurology, 2010, 67, 1252-6.	4.5	53
187	CDKL5 and ARX Mutations in Males With Early-Onset Epilepsy. Pediatric Neurology, 2013, 48, 367-377.	2.1	53
188	Genome-wide enrichment of damaging de novo variants in patients with isolated and complex congenital diaphragmatic hernia. Human Genetics, 2017, 136, 679-691.	3.8	53
189	BRCA1 and BRCA2 mutation carriers in the Breast Cancer Family Registry: an open resource for collaborative research. Breast Cancer Research and Treatment, 2009, 116, 379-386.	2.5	52
190	Combined OXPHOS complex I and IV defect, due to mutated complex I assembly factor C20ORF7. Journal of Inherited Metabolic Disease, 2012, 35, 125-131.	3.6	52
191	The role of genetics in pulmonary arterial hypertension. Journal of Pathology, 2017, 241, 273-280.	4.5	52
192	De novo POGZ mutations are associated with neurodevelopmental disorders and microcephaly. Journal of Physical Education and Sports Management, 2015, 1, a000455.	1.2	51
193	Age-specific breast cancer risk by body mass index and familial risk: prospective family study cohort (ProF-SC). Breast Cancer Research, 2018, 20, 132.	5.0	51
194	Is there a duty to reinterpret genetic data? The ethical dimensions. Genetics in Medicine, 2020, 22, 633-639.	2.4	51
195	A Novel and Lethal De Novo LQT-3 Mutation in a Newborn with Distinct Molecular Pharmacology and Therapeutic Response. PLoS ONE, 2007, 2, e1258.	2.5	50
196	An overview of mongenic and syndromic obesities in humans. Pediatric Blood and Cancer, 2012, 58, 122-128.	1.5	49
197	Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924.	1.1	49
198	Missense variants in the chromatin remodeler CHD1 are associated with neurodevelopmental disability. Journal of Medical Genetics, 2018, 55, 561-566.	3.2	49

#	Article	IF	Citations
199	Increased yield of full GBA sequencing in Ashkenazi Jews with Parkinson's disease. European Journal of Medical Genetics, 2019, 62, 65-69.	1.3	49
200	Biodistribution of onasemnogene abeparvovec DNA, mRNA and SMN protein in human tissue. Nature Medicine, 2021, 27, 1701-1711.	30.7	49
201	New Insights into the Genetics of Fetal Megacystis: ACTG2 Mutations, Encoding γ-2 Smooth Muscle Actin in Megacystis Microcolon Intestinal Hypoperistalsis Syndrome (Berdon Syndrome). Fetal Diagnosis and Therapy, 2015, 38, 296-306.	1.4	48
202	Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635.	3.2	48
203	Characterization of a caveolinâ€1 mutation associated with both pulmonary arterial hypertension and congenital generalized lipodystrophy. Traffic, 2016, 17, 1297-1312.	2.7	48
204	Cohort Profile: The Breast Cancer Prospective Family Study Cohort (ProF-SC). International Journal of Epidemiology, 2016, 45, 683-692.	1.9	48
205	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
206	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
207	Early Pandemic Experiences of Autistic Adults: Predictors of Psychological Distress. Autism Research, 2021, 14, 1209-1219.	3.8	48
208	Considerations Regarding the Genetics of Obesity. Obesity, 2008, 16, S33-9.	3.0	47
209	Common Variants at the 19p13.1 and ZNF365 Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 645-657.	2.5	47
210	Autism Spectrum Disorder, Developmental and Psychiatric Features in 16p11.2 Duplication. Journal of Autism and Developmental Disorders, 2016, 46, 2734-2748.	2.7	47
211	De novo mutations in CSNK2A1 are associated with neurodevelopmental abnormalities and dysmorphic features. Human Genetics, 2016, 135, 699-705.	3.8	47
212	Frequency of GBA Variants in Autopsyâ€proven Multiple System Atrophy. Movement Disorders Clinical Practice, 2017, 4, 574-581.	1.5	47
213	Molecular Genetic Analysis of a Human Neuropeptide Y Receptor. Journal of Biological Chemistry, 1997, 272, 3622-3627.	3.4	46
214	De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	3.2	46
215	Impact of Coronavirus Disease 2019 (COVIDâ€19) on Patients With Congenital Heart Disease Across the Lifespan: The Experience of an Academic Congenital Heart Disease Center in New York City. Journal of the American Heart Association, 2020, 9, e017580.	3.7	46
216	Incidental Findings in the Era of Whole Genome Sequencing?. Hastings Center Report, 2013, 43, 16-19.	1.0	45

#	Article	IF	Citations
217	Weight loss after bariatric surgery in morbidly obese adolescents with MC4R mutations. Obesity, 2014, 22, 225-231.	3.0	45
218	Whole-Exome sequencing identifies novel LEPR mutations in individuals with severe early onset obesity. Obesity, 2014, 22, 576-584.	3.0	45
219	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	2.9	45
220	Identification and characterization of a novel DGAT1 missense mutation associated with congenital diarrhea. Journal of Lipid Research, 2017, 58, 1230-1237.	4.2	44
221	Ethical Considerations Related to Return of Results from Genomic Medicine Projects: The eMERGE Network (Phase III) Experience. Journal of Personalized Medicine, 2018, 8, 2.	2.5	44
222	Regular use of aspirin and other non-steroidal anti-inflammatory drugs and breast cancer risk for women at familial or genetic risk: a cohort study. Breast Cancer Research, 2019, 21, 52.	5.0	44
223	Analysis of Significance Patterns Identifies Ubiquitous and Disease-Specific Gene-Expression Signatures in Patient Peripheral Blood Leukocytes. Annals of the New York Academy of Sciences, 2005, 1062, 146-154.	3.8	43
224	Robust identification of mosaic variants in congenital heart disease. Human Genetics, 2018, 137, 183-193.	3.8	43
225	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
226	Rethinking the â€œopen futureâ€•argument against predictive genetic testing of children. Genetics in Medicine, 2019, 21, 2190-2198.	2.4	43
227	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
228	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. Genome Medicine, 2021, 13, 80.	8.2	43
229	De novo PHIP-predicted deleterious variants are associated with developmental delay, intellectual disability, obesity, and dysmorphic features. Journal of Physical Education and Sports Management, 2016, 2, a001172.	1.2	42
230	Developmental trajectories for young children with 16p11.2 copy number variation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 367-380.	1.7	42
231	Solid phase capturable dideoxynucleotides for multiplex genotyping using mass spectrometry. Nucleic Acids Research, 2002, 30, 85e-85.	14.5	41
232	Effectiveness of a School District Closure for Pandemic Influenza A (H1N1) on Acute Respiratory Illnesses in the Community: A Natural Experiment. Clinical Infectious Diseases, 2013, 56, 509-516.	5.8	41
233	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	3.8	40
234	Genomic Information for Clinicians in the Electronic Health Record: Lessons Learned From the Clinical Genome Resource Project and the Electronic Medical Records and Genomics Network. Frontiers in Genetics, 2019, 10, 1059.	2.3	40

#	Article	IF	Citations
235	SMA-MAP: A Plasma Protein Panel for Spinal Muscular Atrophy. PLoS ONE, 2013, 8, e60113.	2.5	40
236	Molecular physiology of syndromic obesities in humans. Trends in Endocrinology and Metabolism, 2005, 16, 267-272.	7.1	39
237	Research Participantsâ€™ Preferences for Hypothetical Secondary Results from Genomic Research. Journal of Genetic Counseling, 2017, 26, 841-851.	1.6	39
238	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
239	The influence of genetics in congenital diaphragmatic hernia. Seminars in Perinatology, 2020, 44, 151169.	2.5	39
240	Returning Results in the Genomic Era: Initial Experiences of the eMERGE Network. Journal of Personalized Medicine, 2020, 10, 30.	2.5	39
241	16p11.2 deletion syndrome. Current Opinion in Genetics and Development, 2021, 68, 49-56.	3.3	39
242	Genetics of pulmonary hypertension. Current Opinion in Cardiology, 2014, 29, 520-527.	1.8	38
243	Single-Cell Analysis of SMN Reveals Its Broader Role in Neuromuscular Disease. Cell Reports, 2017, 18, 1484-1498.	6.4	38
244	Recessive Rare Variants in Deoxyhypusine Synthase, an Enzyme Involved in the Synthesis of Hypusine, Are Associated with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 287-298.	6.2	38
245	Mutations in ZIC3 and ACVR2B are a common cause of heterotaxy and associated cardiovascular anomalies. Cardiology in the Young, 2012, 22, 194-201.	0.8	37
246	Reciprocal white matter alterations due to 16p11.2 chromosomal deletions versus duplications. Human Brain Mapping, 2016, 37, 2833-2848.	3.6	37
247	Recreational Physical Activity Is Associated with Reduced Breast Cancer Risk in Adult Women at High Risk for Breast Cancer: A Cohort Study of Women Selected for Familial and Genetic Risk. Cancer Research, 2020, 80, 116-125.	0.9	37
248	Comparative Maps of Human 19p13.3 and Mouse Chromosome 10 Allow Identification of Sequences at Evolutionary Breakpoints. Genome Research, 2000, 10, 1369-1380.	5.5	36
249	RAAS gene polymorphisms influence progression of pediatric hypertrophic cardiomyopathy. Human Genetics, 2007, 122, 515-523.	3.8	36
250	Dual Optical Recordings for Action Potentials and Calcium Handling in Induced Pluripotent Stem Cell Models of Cardiac Arrhythmias Using Genetically Encoded Fluorescent Indicators. Stem Cells Translational Medicine, 2015, 4, 468-475.	3.3	36
251	A KCNE2 mutation in a patient with cardiac arrhythmia induced by auditory stimuli and serum electrolyte imbalance. Cardiovascular Research, 2008, 77, 98-106.	3.8	35
252	Association of candidate genes with nonsyndromic clefts in Honduran and Colombian populations. Laryngoscope, 2012, 122, 2082-2087.	2.0	35

#	Article	IF	Citations
253	A Novel LQT-3 Mutation Disrupts an Inactivation Gate Complex with Distinct Rate-Dependent Phenotypic Consequences. Channels, 2007, 1, 273-280.	2.8	34
254	Polymorphism in the Angiotensin II Type 1 Receptor (AGTR1) is Associated With Age at Diagnosis in Pulmonary Arterial Hypertension. Journal of Heart and Lung Transplantation, 2009, 28, 373-379.	0.6	34
255	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	2.5	34
256	A human de novo mutation inMYH10phenocopies the loss of function mutation in mice. Rare Diseases (Austin, Tex), 2013, 1, e26144.	1.8	34
257	The motor neuron response to SMN1 deficiency in spinal muscular atrophy. Muscle and Nerve, 2014, 49, 636-644.	2.2	34
258	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
259	The Impact of Heterozygous KCNK3 Mutations Associated With Pulmonary Arterial Hypertension on Channel Function and Pharmacological Recovery. Journal of the American Heart Association, 2017, 6, .	3.7	34
260	Navigating the researchâ€“clinical interface in genomic medicine: analysis from the CSER Consortium. Genetics in Medicine, 2018, 20, 545-553.	2.4	34
261	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114.	6.2	34
262	Genetic Basis of Human Congenital Heart Disease. Cold Spring Harbor Perspectives in Biology, 2020, 12, a036749.	5.5	34
263	Earlyâ€Onset Parkinsonism Is a Manifestation of the <scp>PPP2R5D</scp> p.<scp>E200K</scp> Mutation. Annals of Neurology, 2020, 88, 1028-1033.	5.3	34
264	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	6.1	34
265	Genotype and defects in microtubule-based motility correlate with clinical severity in KIF1A-associated neurological disorder. Human Genetics and Genomics Advances, 2021, 2, 100026.	1.7	34
266	The mouse mahoganoid coat color mutation disrupts a novel C3HC4 RING domain protein. Journal of Clinical Investigation, 2002, 110, 1449-1459.	8.2	34
267	Novel Association of Early Onset Hepatocellular Carcinoma with Transaldolase Deficiency. JIMD Reports, 2013, 12, 121-127.	1.5	33
268	Report of the National Heart, Lung, and Blood Institute Working Group. Circulation, 2016, 133, 1410-1418.	1.6	33
269	The impact of hereditary cancer gene panels on clinical care and lessons learned. Journal of Physical Education and Sports Management, 2017, 3, a002154.	1.2	33
270	Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. JNCI Cancer Spectrum, 2018, 2, pky023.	2.9	33

#	Article	IF	Citations
271	Psychiatric and Medical Profiles of Autistic Adults in the SPARK Cohort. Journal of Autism and Developmental Disorders, 2020, 50, 3679-3698.	2.7	33
272	Application of ROMA (representational oligonucleotide microarray analysis) to patients with cytogenetic rearrangements. Genetics in Medicine, 2005, 7, 111-118.	2.4	32
273	Correlation of DNA methylation levels in blood and saliva DNA in young girls of the LEGACY Girls study. Epigenetics, 2014, 9, 929-933.	2.7	32
274	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	1.4	32
275	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
276	Global DNA methylation levels in girls with and without a family history of breast cancer. Epigenetics, 2011, 6, 29-33.	2.7	31
277	Intermediate filament protein accumulation in motor neurons derived from giant axonal neuropathy iPSCs rescued by restoration of gigaxonin. Human Molecular Genetics, 2015, 24, 1420-1431.	2.9	31
278	De novo pathogenic variants in CHAMP1 are associated with global developmental delay, intellectual disability, and dysmorphic facial features. Journal of Physical Education and Sports Management, 2016, 2, a000661.	1.2	31
279	Mutations in HIVEP2 are associated with developmental delay, intellectual disability, and dysmorphic features. Neurogenetics, 2016, 17, 159-164.	1.4	31
280	Mutations in TKT Are the Cause of a Syndrome Including Short Stature, Developmental Delay, and Congenital Heart Defects. American Journal of Human Genetics, 2016, 98, 1235-1242.	6.2	31
281	Parental perceptions of prenatal whole exome sequencing (PPPWES) study. Prenatal Diagnosis, 2018, 38, 801-811.	2.3	31
282	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	6.2	31
283	Role of Aberrant Spontaneous Neurotransmission in SNAP25-Associated Encephalopathies. Neuron, 2021, 109, 59-72.e5.	8.1	31
284	Generalizability of Polygenic Risk Scores for Breast Cancer Among Women With European, African, and Latinx Ancestry. JAMA Network Open, 2021, 4, e2119084.	5.9	31
285	GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. ELife, 2020, 9, .	6.0	31
286	Role of a Founder c.201_202delCT Mutation and New Phenotypic Features of Congenital Lipoid Adrenal Hyperplasia in Palestinians. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 4000-4008.	3.6	30
287	Fat discrimination: A phenotype with potential implications for studying fat intake behaviors and obesity. Physiology and Behavior, 2012, 105, 470-475.	2.1	30
288	Differences in Presentation and Outcomes Between Children With Familial Dilated Cardiomyopathy and Children With Idiopathic Dilated Cardiomyopathy. Circulation: Heart Failure, 2017, 10, .	3.9	30

#	Article	IF	Citations
289	Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 14049-14054.	7.1	30
290	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. Journal of the National Cancer Institute, 2019, 111, 350-364.	6.3	30
291	De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. Circulation Genomic and Precision Medicine, 2020, 13, e002836.	3.6	30
292	Knowledge of and Interest in Genetic Results Among Parkinson Disease Patients and Caregivers. Journal of Genetic Counseling, 2014, 23, 114-120.	1.6	29
293	Auditory Evoked M100 Response Latency is Delayed in Children with 16p11.2 Deletion but not 16p11.2 Duplication. Cerebral Cortex, 2016, 26, 1957-1964.	2.9	29
294	Precision Medicine in Children and Young Adults with Hematologic Malignancies and Blood Disorders: The Columbia University Experience. Frontiers in Pediatrics, 2017, 5, 265.	1.9	29
295	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
296	Genetic Causes of Cardiomyopathy in Children: First Results From the Pediatric Cardiomyopathy Genes Study. Journal of the American Heart Association, 2021, 10, e017731.	3.7	29
297	Mild fasting hyperglycemia in children: high rate of glucokinase mutations and some risk of developing type 1 diabetes mellitus. Pediatric Diabetes, 2009, 10, 382-388.	2.9	28
298	Processes and factors involved in decisions regarding return of incidental genomic findings in research. Genetics in Medicine, 2014, 16, 311-317.	2.4	28
299	Loss of function mutation in glutamic pyruvate transaminase 2 (GPT2) causes developmental encephalopathy. Journal of Inherited Metabolic Disease, 2015, 38, 941-948.	3.6	28
300	Professionally Responsible Disclosure of Genomic Sequencing Results in Pediatric Practice. Pediatrics, 2015, 136, e974-e982.	2.1	28
301	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
302	Novel frameshift mutation in Troponin C (TNNC1) associated with hypertrophic cardiomyopathy and sudden death. Cardiology in the Young, 2011, 21, 345-348.	0.8	27
303	Overcoming challenges to meaningful informed consent for whole genome sequencing in pediatric cancer research. Pediatric Blood and Cancer, 2015, 62, 1374-1380.	1.5	27
304	Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829.	3.2	27
305	SARS-CoV-2 Infection in Patients with Down Syndrome, Congenital Heart Disease, and Pulmonary Hypertension: Is Down Syndrome a Risk Factor?. Journal of Pediatrics, 2020, 225, 246-248.	1.8	27
306	Phenotypic expansion of <scp>Boschâ€“Boonstraâ€“Schaaf</scp> optic atrophy syndrome and further evidence for genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2020, 182, 1426-1437.	1.2	27

#	Article	IF	Citations
307	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	4.5	27
308	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. PLoS Genetics, 2020, 16, e1009189.	3.5	27
309	Genetic variants associated with breast cancer risk for Ashkenazi Jewish women with strong family histories but no identifiable BRCA1/2 mutation. Human Genetics, 2013, 132, 523-536.	3.8	26
310	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61.	5.0	26
311	Association of the missense variant p.<scp>Arg203Trp</scp> in <scp>PACS1</scp> as a cause of intellectual disability and seizures. Clinical Genetics, 2017, 92, 221-223.	2.0	26
312	Impact of Receiving Secondary Results from Genomic Research: A 12â€Month Longitudinal Study. Journal of Genetic Counseling, 2018, 27, 709-722.	1.6	26
313	Clinical, biochemical, and molecular overview of transaldolase deficiency and evaluation of the endocrine function: Update of 34 patients. Journal of Inherited Metabolic Disease, 2019, 42, 147-158.	3.6	26
314	A phenome-wide association study to discover pleiotropic effects of PCSK9, APOB, and LDLR. Npj Genomic Medicine, 2019, 4, 3.	3.8	26
315	Second primary breast cancer in BRCA1 and BRCA2 mutation carriers: 10-year cumulative incidence in the Breast Cancer Family Registry. Breast Cancer Research and Treatment, 2015, 151, 653-660.	2.5	25
316	Childhood acromegaly due to X-linked acrogigantism: long term follow-up. Pituitary, 2016, 19, 560-564.	2.9	25
317	Impacts of variants of uncertain significance on parental perceptions of children after prenatal chromosome microarray testing. Prenatal Diagnosis, 2018, 38, 740-747.	2.3	25
318	Participant choices for return of genomic results in the eMERGE Network. Genetics in Medicine, 2020, 22, 1821-1829.	2.4	25
319	United States Pulmonary Hypertension Scientific Registry. Chest, 2021, 159, 311-327.	0.8	25
320	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835.	7.1	25
321	Founder Fukutin mutation causes Walkerâ€“Warburg syndrome in four Ashkenazi Jewish families. Prenatal Diagnosis, 2009, 29, 560-569.	2.3	24
322	Uncovering microdeletions in patients with severe Glut-1 deficiency syndrome using SNP oligonucleotide microarray analysis. Molecular Genetics and Metabolism, 2010, 100, 129-135.	1.1	24
323	Identifying monogenic diabetes in a pediatric cohort with presumed type 1 diabetes. Pediatric Diabetes, 2015, 16, 227-233.	2.9	24
324	Deep Genetic Connection Between Cancer and Developmental Disorders. Human Mutation, 2016, 37, 1042-1050.	2.5	24

#	Article	IF	Citations
325	De novo loss of function mutations in <scp>KIAA2022</scp> are associated with epilepsy and neurodevelopmental delay in females. Clinical Genetics, 2017, 91, 756-763.	2.0	24
326	Predominant and novel de novo variants in 29 individuals with <scp>ALG13</scp> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. Journal of Inherited Metabolic Disease, 2020, 43, 1333-1348.	3.6	24
327	Genetics and Genomics of Pediatric Pulmonary Arterial Hypertension. Genes, 2020, 11, 1213.	2.4	24
328	A Nonsynonymous Polymorphism in IRS1 Modifies Risk of Developing Breast and Ovarian Cancers in BRCA1 and Ovarian Cancer in BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1362-1370.	2.5	23
329	Clinical features of West Nile virus epidemic in Dallas, Texas, 2012. Diagnostic Microbiology and Infectious Disease, 2014, 78, 132-136.	1.8	23
330	Association of a Best-Practice Alert and Prenatal Administration With Tetanus Toxoid, Reduced Diphtheria Toxoid, and Acellular Pertussis Vaccination Rates. Obstetrics and Gynecology, 2015, 126, 333-337.	2.4	23
331	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. Progress in Pediatric Cardiology, 2016, 40, 41-45.	0.4	23
332	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	2.5	23
333	Evaluating heterogeneity in <scp>ASD</scp> symptomatology, cognitive ability, and adaptive functioning among 16p11.2 <scp>CNV</scp> carriers. Autism Research, 2020, 13, 1300-1310.	3.8	23
334	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	2.4	23
335	A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. New England Journal of Medicine, 2021, 385, 1292-1301.	27.0	23
336	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
337	Developments in Molecular Genetic Diagnostics: An Update for the Pediatric Epilepsy Specialist. Pediatric Neurology, 2011, 44, 317-327.	2.1	22
338	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	2.5	22
339	Applying Deep Neural Network Analysis to High-Content Image-Based Assays. SLAS Discovery, 2019, 24, 829-841.	2.7	22
340	Comparative outcomes of right versus left congenital diaphragmatic hernia: A multicenter analysis. Journal of Pediatric Surgery, 2020, 55, 33-38.	1.6	22
341	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
342	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22

#	Article	IF	Citations
343	Biallelic variants of ATP13A3 cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. Journal of Medical Genetics, 2022, 59, 906-911.	3.2	22
344	Rare and de novo variants in 827 congenital diaphragmatic hernia probands implicate LONP1 as candidate risk gene. American Journal of Human Genetics, 2021, 108, 1964-1980.	6.2	22
345	Variants of the CFC1 gene in patients with laterality defects associated with congenital cardiac disease. Cardiology in the Young, 2007, 17, 268-274.	0.8	21
346	Implementation of Genetics to Personalize Medicine. Gender Medicine, 2007, 4, 248-265.	1.4	21
347	Views of internists towards uses of PGD. Reproductive BioMedicine Online, 2013, 26, 142-147.	2.4	21
348	Common germline-somatic variant interactions in advanced urothelial cancer. Nature Communications, 2020, 11, 6195.	12.8	21
349	Likely damaging de novo variants in congenital diaphragmatic hernia patients are associated with worse clinical outcomes. Genetics in Medicine, 2020, 22, 2020-2028.	2.4	21
350	Developmental basis of trachea-esophageal birth defects. Developmental Biology, 2021, 477, 85-97.	2.0	21
351	The genetic architecture of pediatric cardiomyopathy. American Journal of Human Genetics, 2022, 109, 282-298.	6.2	21
352	Familial X-Linked Acrogigantism: Postnatal Outcomes and Tumor Pathology in a Prenatally Diagnosed Infant and His Mother. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4667-4675.	3.6	20
353	Genetics and Other Omics in Pediatric Pulmonary Arterial Hypertension. Chest, 2020, 157, 1287-1295.	0.8	20
354	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
355	Association of Allelic Variation in Genes Mediating Aspects of Energy Homeostasis with Weight Gain during Administration of Antipsychotic Drugs (CATIE Study). Frontiers in Genetics, 2011, 2, 56.	2.3	19
356	Abnormal auditory and language pathways in children with 16p11.2 deletion. NeuroImage: Clinical, 2015, 9, 50-57.	2.7	19
357	Relationship between M100 Auditory Evoked Response and Auditory Radiation Microstructure in 16p11.2 Deletion and Duplication Carriers. American Journal of Neuroradiology, 2016, 37, 1178-1184.	2.4	19
358	Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478.	1.2	19
359	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2019, 121, 180-192.	6.4	19
360	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	2.8	19

#	Article	IF	Citations
361	Rapid exome sequencing in PICU patients with new-onset metabolic or neurological disorders. Pediatric Research, 2020, 88, 761-768.	2.3	19
362	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	12.8	19
363	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
364	Post-translational formation of hypusine in eIF5A: implications in human neurodevelopment. Amino Acids, 2022, 54, 485-499.	2.7	19
365	Neurogenetic disorders across the lifespan: from aberrant development to degeneration. Nature Reviews Neurology, 2022, 18, 117-124.	10.1	19
366	Loss-of-function variants in TIAM1 are associated with developmental delay, intellectual disability, and seizures. American Journal of Human Genetics, 2022, 109, 571-586.	6.2	19
367	Oligonucleotide array CGH studies in myeloproliferative neoplasms: Comparison with JAK2V617F mutational status and conventional chromosome analysis. Leukemia Research, 2009, 33, 662-664.	0.8	18
368	Psychiatristsâ€™ Views of the Genetic Bases of Mental Disorders and Behavioral Traits and Their Use of Genetic Tests. Journal of Nervous and Mental Disease, 2014, 202, 530-538.	1.0	18
369	Genetic causal attribution of epilepsy and its implications for felt stigma. Epilepsia, 2015, 56, 1542-1550.	5.1	18
370	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134.	2.5	18
371	Harmonizing Outcomes for Genomic Medicine: Comparison of eMERGE Outcomes to ClinGen Outcome/Intervention Pairs. Healthcare (Switzerland), 2018, 6, 83.	2.0	18
372	Clinical presentation and natural history of infantile-onset ascending spastic paralysis from three families with an ALS2 founder variant. Neurological Sciences, 2018, 39, 1917-1925.	1.9	18
373	Impact of patient education videos on genetic counseling outcomes after exome sequencing. Patient Education and Counseling, 2020, 103, 127-135.	2.2	18
374	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	6.2	18
375	Neurodevelopmental phenotypes associated with pathogenic variants in SLC6A1. Journal of Medical Genetics, 2022, 59, 536-543.	3.2	18
376	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 1330-1341.	6.2	18
377	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study. Circulation, 2022, 145, 877-891.	1.6	18
378	Preimplantation genetic diagnosis on in vitro fertilization clinic websites: presentations of risks, benefits and other information. Fertility and Sterility, 2009, 92, 1276-1283.	1.0	17

#	Article	IF	Citations
379	Novel loci interacting epistatically with bone morphogenetic protein receptor 2 cause familial pulmonary arterial hypertension. Journal of Heart and Lung Transplantation, 2010, 29, 174-180.	0.6	17
380	The Expanding MEGDEL Phenotype: Optic Nerve Atrophy, Microcephaly, and Myoclonic Epilepsy in a Child with SERAC1 Mutations. JIMD Reports, 2014, 16, 75-79.	1.5	17
381	Genetic Testing of Children for Diseases That Have Onset in Adulthood: The Limits of Family Interests. Pediatrics, 2014, 134, S104-S110.	2.1	17
382	Association of Researcher Characteristics with Views on Return of Incidental Findings from Genomic Research. Journal of Genetic Counseling, 2015, 24, 833-841.	1.6	17
383	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American Journal of Human Genetics, 2018, 103, 786-793.	6.2	17
384	COVID-19â€™s Impact on Genetics at One Medical Center in New York. Genetics in Medicine, 2020, 22, 1467-1469.	2.4	17
385	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130.	2.8	17
386	Heterozygous variants that disturb the transcriptional repressor activity of FOXP4 cause a developmental disorder with speech/language delays and multiple congenital abnormalities. Genetics in Medicine, 2021, 23, 534-542.	2.4	17
387	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
388	Polymerase Chain Reactionâ€Restriction Fragment Length Polymorphisms (PCRâ€RFLP) and Electrophoretic Assays for the Mouse Obese (Lep^{ob}) Mutation. Obesity, 1997, 5, 183-185.	4.0	16
389	Analysis of 30 Genes (355 SNPS) Related to Energy Homeostasis for Association with Adiposity in European-American and Yupâ€™ik Eskimo Populations. Human Heredity, 2009, 67, 193-205.	0.8	16
390	Recipient Genotype Is a Predictor of Allograft Cytokine Expression and Outcomes After Pediatric Cardiac Transplantation. Journal of the American College of Cardiology, 2009, 53, 1909-1917.	2.8	16
391	Genetic testing preferences in families containing multiple individuals with epilepsy. Epilepsia, 2014, 55, 1705-1713.	5.1	16
392	Researchersâ€™ views on informed consent for return of secondary results in genomic research. Genetics in Medicine, 2015, 17, 644-650.	2.4	16
393	Examining the Psychosocial Impact of Genetic Testing for Cardiomyopathies. Journal of Genetic Counseling, 2018, 27, 927-934.	1.6	16
394	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	1.4	16
395	Language characterization in 16p11.2 deletion and duplication syndromes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 380-391.	1.7	16
396	A qualitative study of Latinx parents' experiences of clinical exome sequencing. Journal of Genetic Counseling, 2020, 29, 574-586.	1.6	16

#	Article	IF	Citations
397	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq1 1 0.784314 rgBT /Overlock	2.5	16
398	Genes that drive the pathobiology of pediatric pulmonary arterial hypertension. Pediatric Pulmonology, 2021, 56, 614-620.	2.0	16
399	Detailed Clinical and Psychological Phenotype of the X-linked HNRNPH2-Related Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e551.	1.9	16
400	Availability of Services and Caregiver Burden: Supporting Individuals With Neurogenetic Conditions During the COVID-19 Pandemic. Journal of Child Neurology, 2021, 36, 760-767.	1.4	16
401	The predictive ability of the 313 variantâ€“based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. Genetics in Medicine, 2021, 23, 1726-1737.	2.4	16
402	GeneLiFT: A novel test to facilitate rapid screening of genetic literacy in a diverse population undergoing genetic testing. Journal of Genetic Counseling, 2021, 30, 742-754.	1.6	16
403	Oligonucleotide Array CGH Studies in Myeloproliferative Neoplasms and Comparison with Conventional Cytogenetic Analysis. Blood, 2007, 110, 1550-1550.	1.4	16
404	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
405	Molecular Mapping of the Tubby (tub) Mutation on Mouse Chromosome 7. Genomics, 1996, 32, 210-217.	2.9	15
406	A unique case of der(11)t(11;22),-22 arising from 3:1 segregation of a maternal t(11;22) in a family with co-segregation of the translocation and breast cancer. Prenatal Diagnosis, 2005, 25, 683-686.	2.3	15
407	Clinical Application of Whole-Exome Sequencing. JAMA Neurology, 2013, 70, 788.	9.0	15
408	Approaches to carrier testing and results disclosure in translational genomics research: The clinical sequencing exploratory research consortium experience. Molecular Genetics & Genomic Medicine, 2018, 6, 898-909.	1.2	15
409	Glucokinase mutations in young children with hyperglycemia. Diabetes/Metabolism Research and Reviews, 2006, 22, 348-355.	4.0	14
410	The effect of cardiac genetic testing on psychological well-being and illness perceptions. Heart and Lung: Journal of Acute and Critical Care, 2014, 43, 127-132.	1.6	14
411	Clinical and genetic characterization of individuals with predicted deleterious PHIP variants. Journal of Physical Education and Sports Management, 2019, 5, a004200.	1.2	14
412	Increasing genomic literacy among adolescents. Genetics in Medicine, 2019, 21, 994-1000.	2.4	14
413	Prepubertal Internalizing Symptoms and Timing of Puberty Onset in Girls. American Journal of Epidemiology, 2021, 190, 431-438.	3.4	14
414	Does the law require reinterpretation and return of revised genomic results?. Genetics in Medicine, 2021, 23, 833-836.	2.4	14

#	Article	IF	Citations
415	Penetrance of Breast Cancer Susceptibility Genes from the eMERGE III Network. JNCI Cancer Spectrum, 2021, 5, pkab044.	2.9	14
416	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.7	14
417	Prenatal diagnosis of congenital lipoid adrenal hyperplasia (CLAH) by estriol amniotic fluid analysis and molecular genetic testing. Prenatal Diagnosis, 2008, 28, 11-14.	2.3	13
418	Comparison of Endoscopic and Clinical Characteristics of Patients with Familial and Sporadic Barrettâ€™s Esophagus. Digestive Diseases and Sciences, 2011, 56, 1702-1706.	2.3	13
419	Psychosocial Adjustment in School-age Girls With a Family History of Breast Cancer. Pediatrics, 2015, 136, 927-937.	2.1	13
420	Population-Based Study of Attitudes towardBRCAGenetic Testing among Orthodox Jewish Women. Breast Journal, 2017, 23, 333-337.	1.0	13
421	23andMe Paves the Way for Direct-to-Consumer Genetic Health Risk Tests of Limited Clinical Utility. Annals of Internal Medicine, 2017, 167, 125.	3.9	13
422	Pulmonary hypertension in patients with 9q34.3 microdeletionâ€associated Kleefstra syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1773-1777.	1.2	13
423	Cases in Precision Medicine: The Role of Polygenic Risk Scores in Breast Cancer Risk Assessment. Annals of Internal Medicine, 2021, 174, 408-412.	3.9	13
424	Comparing 5-Year and Lifetime Risks of Breast CancerÂusing the Prospective Family Study Cohort. Journal of the National Cancer Institute, 2021, 113, 785-791.	6.3	13
425	Is there a way to reduce the inequity in variant interpretation on the basis of ancestry?. American Journal of Human Genetics, 2022, 109, 981-988.	6.2	13
426	Eating in the absence of hunger but not loss of control behaviors are associated with 16p11.2 deletions. Obesity, 2014, 22, 2625-2631.	3.0	12
427	Pulmonary arterial hypertension: Specialistsâ€™ knowledge, practices, and attitudes of genetic counseling and genetic testing in the USA. Pulmonary Circulation, 2017, 7, 372-383.	1.7	12
428	Tandem mass spectrometry assay of Î²-glucocerebrosidase activity in dried blood spots eliminates false positives detected in fluorescence assay. Molecular Genetics and Metabolism, 2018, 123, 135-139.	1.1	12
429	Precision Medicine in Internal Medicine. Annals of Internal Medicine, 2019, 170, 635.	3.9	12
430	The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network. Genetics in Medicine, 2022, 24, 1130-1138.	2.4	12
431	The Links Between Obesity, Leptin, and Prostate Cancer. Cancer Journal (Sudbury, Mass), 2006, 12, 178-181.	2.0	11
432	Two Cases of Pulmonary Hypertension Associated with Type III Glycogen Storage Disease. JIMD Reports, 2011, 1, 79-82.	1.5	11

#	Article	IF	Citations
433	Should Life Insurers Have Access to Genetic Test Results?. JAMA - Journal of the American Medical Association, 2014, 312, 1855.	7.4	11
434	Recurrent diffuse lung disease due to surfactant protein C deficiency. Respiratory Medicine Case Reports, 2018, 25, 91-95.	0.4	11
435	Functional Consequences of the SCN5A-p.Y1977N Mutation within the PY Ubiquitylation Motif: Discrepancy between HEK293 Cells and Transgenic Mice. International Journal of Molecular Sciences, 2019, 20, 5033.	4.1	11
436	Sensorimotor Cortical Oscillations during Movement Preparation in 16p11.2 Deletion Carriers. Journal of Neuroscience, 2019, 39, 7321-7331.	3.6	11
437	Psychotic symptoms in 16p11.2 copyâ€number variant carriers. Autism Research, 2020, 13, 187-198.	3.8	11
438	Differences in brain structure and function in children with the FTO obesityâ€risk allele. Obesity Science and Practice, 2020, 6, 409-424.	1.9	11
439	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
440	Novel SLC39A4 Mutation in Acrodermatitis Enteropathica. Pediatric Dermatology, 2011, 28, 697-700.	0.9	10
441	Determinants of extracellular matrix remodelling are differentially expressed in paediatric and adult dilated cardiomyopathy. European Journal of Heart Failure, 2011, 13, 271-277.	7.1	10
442	Breast cancer family history and allele-specific DNA methylation in the legacy girls study. Epigenetics, 2018, 13, 240-250.	2.7	10
443	Assessing patient readiness for personalized genomic medicine. Journal of Community Genetics, 2019, 10, 109-120.	1.2	10
444	Biallelic variants in AGMO with diminished enzyme activity are associated with a neurodevelopmental disorder. Human Genetics, 2019, 138, 1259-1266.	3.8	10
445	Evaluation of the cost and effectiveness of diverse recruitment methods for a genetic screening study. Genetics in Medicine, 2019, 21, 2371-2380.	2.4	10
446	Cases in Precision Medicine: The Role of Tumor and Germline Genetic Testing in Breast Cancer Management. Annals of Internal Medicine, 2019, 171, 925.	3.9	10
447	A novel homozygous variant in TRAPPC2L results in a neurodevelopmental disorder and disrupts TRAPP complex function. Journal of Medical Genetics, 2021, 58, 592-601.	3.2	10
448	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
449	Weightâ€loss response to naltrexone/bupropion is modulated by the <scp>Taq1A</scp> genetic variant near <scp>DRD2</scp> (<scp>rs1800497</scp>): A pilot study. Diabetes, Obesity and Metabolism, 2021, 23, 850-853.	4.4	10
450	Views of preimplantation genetic diagnosis among psychiatrists and neurologists. Journal of reproductive medicine, The, 2014, 59, 385-92.	0.2	10

#	Article	IF	Citations
451	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. Clinical Epigenetics, 2022, 14, 52.	4.1	10
452	Newborn screening for neurodevelopmental diseases: Are we there yet?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 222-230.	1.6	10
453	Alpha-Thalassemia Major Presenting in a Term Neonate without Hydrops. Pediatric and Developmental Pathology, 2005, 8, 706-709.	1.0	9
454	When to Offer Genetic Testing for Pulmonary Arterial Hypertension. Canadian Journal of Cardiology, 2015, 31, 544-547.	1.7	9
455	A Quality Improvement Collaborative to Improve Pediatric Primary Care Genetic Services. Pediatrics, 2016, 137, e20143874.	2.1	9
456	Benign breast disease increases breast cancer risk independent of underlying familial risk profile: Findings from a Prospective Family Study Cohort. International Journal of Cancer, 2019, 145, 370-379.	5.1	9
457	Frequency and characterization of mutations in genes in a large cohort of patients referred to MODY registry. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 633-638.	0.9	9
458	Neurodevelopmental phenotypes in individuals with pathogenic variants in CHAMP1. Journal of Physical Education and Sports Management, 2021, 7, a006092.	1.2	9
459	The association between congenital diaphragmatic hernia and undescended testes. Journal of Pediatric Surgery, 2015, 50, 744-745.	1.6	8
460	Whole-Exome Sequencing Reveals CLCNKB Mutations in a Case of Sudden Unexpected Infant Death. Pediatric and Developmental Pathology, 2015, 18, 324-326.	1.0	8
461	Adrenergic receptor genotype influences heart failure severity and Î²-blocker response in children with dilated cardiomyopathy. Pediatric Research, 2015, 77, 363-369.	2.3	8
462	Parentsâ€™ interest in genetic testing of their offspring in multiplex epilepsy families. Epilepsia, 2016, 57, 279-287.	5.1	8
463	Accuracy of Risk Estimates from the iPrevent Breast Cancer Risk Assessment and Management Tool. JNCI Cancer Spectrum, 2019, 3, pkz066.	2.9	8
464	Cases in Precision Medicine: The Role of Pharmacogenetics in Precision Prescribing. Annals of Internal Medicine, 2019, 170, 796.	3.9	8
465	De novo heterozygous missense and lossâ€ofâ€function variants in CDC42BPB are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973.	1.2	8
466	The Steroid Metabolome and Breast Cancer Risk in Women with a Family History of Breast Cancer: The Novel Role of Adrenal Androgens and Glucocorticoids. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 89-96.	2.5	8
467	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	2.4	8
468	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003500.	3.6	8

#	Article	IF	Citations
469	Predictive genetic testing for cardiomyopathies. Progress in Pediatric Cardiology, 2007, 23, 33-38.	0.4	7
470	Hypertrophic cardiomyopathy: New approaches and a time to reappraise older approaches. Journal of Thoracic and Cardiovascular Surgery, 2016, 152, 983-988.	0.8	7
471	Depression and genetic causal attribution of epilepsy in multiplex epilepsy families. Epilepsia, 2016, 57, 1643-1650.	5.1	7
472	A definition of gentle ventilation in congenital diaphragmatic hernia: a survey of neonatologists and pediatric surgeons. Journal of Perinatal Medicine, 2017, 45, 1031-1038.	1.4	7
473	Biallelic variants in VARS in a family with two siblings with intellectual disability and microcephaly: case report and review of the literature. Journal of Physical Education and Sports Management, 2018, 4, a003301.	1.2	7
474	Laboratory considerations for prenatal genetic testing. Seminars in Perinatology, 2018, 42, 307-313.	2.5	7
475	United States Pulmonary Hypertension Scientific Registry (USPHSR): rationale, design, and clinical implications. Pulmonary Circulation, 2019, 9, 204589401985169.	1.7	7
476	Association of Prepubertal and Adolescent Androgen Concentrations With Timing of Breast Development and Family History of Breast Cancer. JAMA Network Open, 2019, 2, e190083.	5.9	7
477	Cases in Precision Medicine: When Patients Present With Direct-to-Consumer Genetic Test Results. Annals of Internal Medicine, 2019, 170, 643.	3.9	7
478	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	2.0	7
479	Variants in STXBP3 are Associated with Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation. Journal of Crohn's and Colitis, 2021, 15, 1908-1919.	1.3	7
480	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
481	Dilated cardiomyopathy due to a phospholamban duplication. Cardiology in the Young, 2014, 24, 953-954.	0.8	6
482	Features of Feingold syndrome 1 dominate in subjects with 2p deletions including MYCN. American Journal of Medical Genetics, Part A, 2018, 176, 1956-1963.	1.2	6
483	A GENETIC FIRST APPROACH TO DISSECTING THE HETEROGENEITY OF AUTISM: PHENOTYPIC COMPARISON OF AUTISM RISK COPY NUMBER VARIANTS. European Neuropsychopharmacology, 2019, 29, S783-S784.	0.7	6
484	Dallas MegaShelter Medical Operations Response to Hurricane Harvey. Disaster Medicine and Public Health Preparedness, 2019, 13, 90-93.	1.3	6
485	Human iPSC-Derived Neuronal Cells From CTBP1-Mutated Patients Reveal Altered Expression of Neurodevelopmental Gene Networks. Frontiers in Neuroscience, 2020, 14, 562292.	2.8	6
486	The psychiatric phenotypes of 1q21 distal deletion and duplication. Translational Psychiatry, 2021, 11, 105.	4.8	6

#	Article	IF	Citations
487	Gene expression atlas of energy balance brain regions. JCI Insight, 2021, 6, .	5.0	6
488	Detecting potential pleiotropy across cardiovascular and neurological diseases using univariate, bivariate, and multivariate methods on 43,870 individuals from the eMERGE network. , 2018, , .		6
489	Cross-sectional, quantitative analysis of motor function in females with HNRNPH2-related disorder. Research in Developmental Disabilities, 2021, 119, 104110.	2.2	6
490	Genotype-Phenotype Comparison in POGZ-Related Neurodevelopmental Disorders by Using Clinical Scoring. Genes, 2022, 13, 154.	2.4	6
491	Channelopathy Genes in Pulmonary Arterial Hypertension. Biomolecules, 2022, 12, 265.	4.0	6
492	Clinical and Molecular Genetic Features of Hereditary Pulmonary Arterial Hypertension. , 2011, 1, 1721-1728.		5
493	Intragenic deletion as a novel type of mutation in Wolman disease. Molecular Genetics and Metabolism, 2011, 104, 703-705.	1.1	5
494	Human Subjects Protection: An Event Monitoring Committee for Research Studies of Girls From Breast Cancer Families. Journal of Adolescent Health, 2014, 55, 352-357.	2.5	5
495	Rapidly progressive mitral valve stenosis in patients with acromelic dysplasia. Cardiology in the Young, 2017, 27, 797-800.	0.8	5
496	Whole-Genome and Whole-Exome Sequencing in Pediatric Oncology: An Assessment of Parent and Young Adult Patient Knowledge, Attitudes, and Expectations. JCO Precision Oncology, 2018, 2, 1-11.	3.0	5
497	Influence of pubertal development on urinary oxidative stress biomarkers in adolescent girls in the New York LEGACY cohort. Free Radical Research, 2020, 54, 431-441.	3.3	5
498	Deep whole-genome sequencing of multiple proband tissues and parental blood reveals the complex genetic etiology of congenital diaphragmatic hernias. Human Genetics and Genomics Advances, 2020, 1, 100008.	1.7	5
499	Returning negative results from <scp>largeâ€scale</scp> genomic screening: Experiences from the <scp>eMERGE III</scp> network. American Journal of Medical Genetics, Part A, 2021, 185, 508-516.	1.2	5
500	Genetic Variant Reinterpretation: Economic and Population Health Management Challenges. Population Health Management, 2021, 24, 310-313.	1.7	5
501	An electronic health record (EHR) log analysis shows limited clinician engagement with unsolicited genetic test results. JAMIA Open, 2021, 4, ooab014.	2.0	5
502	Response to Li and Hopper. American Journal of Human Genetics, 2021, 108, 527-529.	6.2	5
503	Reproductive decisionâ€making in families containing multiple individuals with epilepsy. Epilepsia, 2021, 62, 1220-1230.	5.1	5
504	Recommendation of premarital genetic screening in the Syrian Jewish community based on mutation carrier frequencies within Syrian Jewish cohorts. Molecular Genetics & Genomic Medicine, 2021, 9, e1756.	1.2	5

#	Article	IF	Citations
505	Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders. Journal of Medical Genetics, 2022, 59, 669-677.	3.2	5
506	Neuropathological Findings in a Case of Parkinsonism and Developmental Delay Associated with a Monoallelic Variant in <scp>PLXNA1</scp>. Movement Disorders, 2021, 36, 2681-2687.	3.9	5
507	Powerful gene-based testing by integrating long-range chromatin interactions and knockoff genotypes. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	5
508	Genetics Dictating Therapeutic Decisions in Pediatric Pulmonary Hypertension? A Case Report Suggesting We Are Getting Closer.. Pulmonary Circulation, 2022, 12, e12033.	1.7	5
509	Neither cardiac mitochondrial DNA variation nor copy number contribute to congenital heart disease risk. American Journal of Human Genetics, 2022, 109, 961-966.	6.2	5
510	Foamy podocytes. American Journal of Kidney Diseases, 2003, 41, 891-896.	1.9	4
511	Novel gene discovery in pediatric cardiomyopathy. Progress in Pediatric Cardiology, 2011, 31, 89-91.	0.4	4
512	Evaluation of the <scp>CAV</scp>1 gene in clinically, sonographically and histologically proven morphea patients. Experimental Dermatology, 2015, 24, 718-720.	2.9	4
513	How to effectively utilize genetic testing in the care of children with cardiomyopathies. Progress in Pediatric Cardiology, 2015, 39, 3-11.	0.4	4
514	Nonspecific phenotype of Noonan syndrome diagnosed by whole exome sequencing. Clinical Case Reports (discontinued), 2015, 3, 237-239.	0.5	4
515	Understanding Factors Associated with Uptake of<i> BRCA1/2</i> Genetic Testing<i></i> among Orthodox Jewish Women in the USA Using a Mixed-Methods Approach. Public Health Genomics, 2018, 21, 186-196.	1.0	4
516	Genetic attribution and perceived impact of epilepsy in multiplex epilepsy families. Epilepsia, 2019, 60, 2286-2293.	5.1	4
517	Homozygous noncanonical splice variant in LSM1 in two siblings with multiple congenital anomalies and global developmental delay. Journal of Physical Education and Sports Management, 2019, 5, a004101.	1.2	4
518	Choices, attitudes, and experiences of genetic screening in Latino/a and Ashkenazi Jewish individuals. Journal of Community Genetics, 2020, 11, 391-403.	1.2	4
519	Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes. Genome Medicine, 2021, 13, 147.	8.2	4
520	Extracorporeal membrane oxygenation (ECMO) and its complications in newborns with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2022, , .	1.6	4
521	Genetic issues in pediatric cardiomyopathy: Future research directions. Progress in Pediatric Cardiology, 2011, 32, 3-4.	0.4	3
522	Mutations in BMPR2 are not present in patients with pulmonary hypertension associated with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2017, 52, 1747-1750.	1.6	3

#	Article	IF	Citations
523	Selfâ€Reported Questionnaire Detects Family History of Cancer in a Pancreatic Cancer Screening Program. Journal of Genetic Counseling, 2017, 26, 806-813.	1.6	3
524	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. Human Mutation, 2018, 39, 870-881.	2.5	3
525	Genetics of pediatric cardiomyopathies. Progress in Pediatric Cardiology, 2018, 49, 18-19.	0.4	3
526	Common Childhood Viruses and Pubertal Timing: The LEGACY Girls Study. American Journal of Epidemiology, 2021, 190, 766-778.	3.4	3
527	Clinical and genomic characterization of 8p cytogenomic disorders. Genetics in Medicine, 2021, 23, 2342-2351.	2.4	3
528	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	2.4	3
529	Case Report: Esophageal Bronchus in a Neonate, With Image, Histological, and Molecular Analysis. Frontiers in Pediatrics, 2021, 9, 707822.	1.9	3
530	Biâ€allelic PAGR1 variants are associated with microcephaly and a severe neurodevelopmental disorder: Genetic evidence from two families. American Journal of Medical Genetics, Part A, 2021, , .	1.2	3
531	Reimbursement for genetic variant reinterpretation: five questions payers should ask. American Journal of Managed Care, 2021, 27, e336-e338.	1.1	3
532	Neurodevelopmental profile of HIVEP2â€related disorder. Developmental Medicine and Child Neurology, 2022, 64, 654-661.	2.1	3
533	Abnormal Vertical Eye Movements as a Clue for Diagnosis of Niemann-Pick Type C. Tremor and Other Hyperkinetic Movements, 2018, 8, 560.	2.0	3
534	Information is power: The experiences, attitudes and needs of individuals who chose to have prenatal genomic sequencing for fetal anomalies. Prenatal Diagnosis, 2022, 42, 947-954.	2.3	3
535	Delineation of New Disorders and Phenotypic Expansion of Known Disorders Through Whole Exome Sequencing. Current Genetic Medicine Reports, 2015, 3, 209-218.	1.9	2
536	ACSS2 gene variant associated with cleft lip and palate in two independent Hispanic populations. Laryngoscope, 2017, 127, E336-E339.	2.0	2
537	KCNJ11 Mutation in One Family is Associated with Adult-Onset Rather than Neonatal-Onset Diabetes Mellitus. AACE Clinical Case Reports, 2018, 4, e411-e414.	1.1	2
538	Response to ten Broeke et al.. Genetics in Medicine, 2019, 21, 258-259.	2.4	2
539	Identification of a secondary RET mutation in a pediatric patient with relapsed acute myeloid leukemia leads to the diagnosis and treatment of asymptomatic metastatic medullary thyroid cancer in a parent: a case for sequencing the germline. Journal of Physical Education and Sports Management, 2019, 5, a003889.	1.2	2
540	Cases in Precision Medicine: Genetic Assessment After a Sudden Cardiac Death in the Family. Annals of Internal Medicine, 2019, 170, 710.	3.9	2

#	Article	IF	Citations
541	Genomic medicine implementation protocols in the PhenX Toolkit: tools for standardized data collection. Genetics in Medicine, 2021, 23, 1783-1788.	2.4	2
542	Impact of Genetic Testing for Cardiomyopathy on Emotional Well-Being and Family Dynamics: A Study of Parents and Adolescents. Circulation Genomic and Precision Medicine, 2021, 14, e003189.	3.6	2
543	Harnessing rare variants in neuropsychiatric and neurodevelopment disordersâ€"a Keystone Symposia report. Annals of the New York Academy of Sciences, 2021, , .	3.8	2
544	iPSC-derived Î² cells model diabetes due to glucokinase deficiency. Journal of Clinical Investigation, 2017, 127, 1115-1115.	8.2	2
545	Association Between Genetic Testing for Hereditary Breast Cancer and Contralateral Prophylactic Mastectomy Among Multiethnic Women Diagnosed With Early-Stage Breast Cancer. JCO Oncology Practice, 2021, , OP2100322.	2.9	2
546	Identification and validation of candidate risk genes in endocytic vesicular trafficking associated with esophageal atresia and tracheoesophageal fistulas. Human Genetics and Genomics Advances, 2022, 3, 100107.	1.7	2
547	Catecholaminergic polymorphic ventricular tachycardia in a child with Brugada pattern on ECG: One patient with two diseases?. Heart Rhythm, 2014, 11, 2101-2104.	0.7	1
548	Future research directions in pediatric cardiomyopathy. Progress in Pediatric Cardiology, 2016, 40, 35-39.	0.4	1
549	Clinical, biochemical, and molecular overview of transaldolase deficiency and evaluation of the endocrine function: update of 34 patients. Journal of Inherited Metabolic Disease, 2019, 42, 147.	3.6	1
550	Genomic Sequencing for Infants and Children in Intensive Care Units. Current Pediatrics Reports, 2019, 7, 78-82.	4.0	1
551	Response to Evans et al.. Genetics in Medicine, 2019, 21, 1880-1881.	2.4	1
552	Abnormal Auditory Mismatch Fields in Children and Adolescents With 16p11.2 Deletion and 16p11.2 Duplication. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2020, 5, 942-950.	1.5	1
553	Reply to â€œ<scp>PPP2R5D</scp> Genetic Mutations and Early Onset Parkinsonismâ€• Annals of Neurology, 2021, 89, 195-196.	5.3	1
554	Practice Patterns After Return of Rare Variants Associated With Cardiomyopathy in the Electronic Medical Records and Genomics Network. Circulation: Heart Failure, 2021, 14, e008155.	3.9	1
555	Implementation of Population-Based Newborn Screening Reveals Low Incidence of Spinal Muscular Atrophy. Obstetrical and Gynecological Survey, 2021, 76, 17-19.	0.4	1
556	Utility of Oligonucleotide Array Comparative Genomic Hybridization to Identify Cryptic Copy Number Alterations in Myelodysplastic Syndromes. Blood, 2008, 112, 5076-5076.	1.4	1
557	OP035: Rapid Whole Genome Sequencing (rWGS) in the cardiac NICU. Genetics in Medicine, 2022, 24, S362-S363.	2.4	1
558	Generation of three induced pluripotent stem cells lines from patients with esophageal atresia/tracheoesophageal fistula type C. Stem Cell Research, 2022, 60, 102711.	0.7	1

#	Article	IF	Citations
559	Recreational Physical Activity and Outcomes After Breast Cancer in Women at High Familial Risk. JNCI Cancer Spectrum, 2021, 5, pkab090.	2.9	1
560	A homozygous splice variant in ATP5PO , disrupts mitochondrial complex V function and causes Leigh syndrome in two unrelated families. Journal of Inherited Metabolic Disease, 0, , .	3.6	1
561	Women's thoughts on receiving and sharing genetic information: Considerations for genetic counseling. Journal of Genetic Counseling, 0, , .	1.6	1
562	Blood leukocyte microarrays to diagnose systemic onset juvenile idiopathic arthritis and follow the response to IL-1 blockade. Journal of Experimental Medicine, 2009, 206, 2299-2299.	8.5	0
563	Are KCNK3 Mutations Found in Vasoreactive Patients With Idiopathic Pulmonary Arterial Hypertension?. Chest, 2013, 144, 878A.	0.8	0
564	Reply. Muscle and Nerve, 2014, 50, 458-459.	2.2	0
565	KCNK3 Mutations Are Found in Patients With Pulmonary Arterial Hypertension Associated With Fenfluramine or Dexfenfluramine Ingestion. Chest, 2015, 148, 944A.	0.8	0
566	Genetics and Hypertrophic Cardiomyopathy. Current Pediatrics Reports, 2016, 4, 35-44.	4.0	0
567	IMPACT OF DAMAGING DE NOVO VARIANTS ON CLINICAL OUTCOMES IN CONGENITAL HEART DISEASE. Journal of the American College of Cardiology, 2017, 69, 2553.	2.8	0
568	Analysis of myocardial fibrosis in children with hypertrophic cardiomyopathy: A report from the pediatric cardiomyopathy registry. Progress in Pediatric Cardiology, 2017, 46, 33.	0.4	0
569	Response to Dr. Sorscher. Journal of Genetic Counseling, 2017, 26, 1164-1164.	1.6	0
570	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
571	Response to Wang et al.. Genetics in Medicine, 2019, 21, 2158.	2.4	0
572	Cases in Precision Medicine: Should You Participate in a Study Involving Genomic Sequencing of Your Patients?. Annals of Internal Medicine, 2019, 171, 568.	3.9	0
573	T148. Structural and Functional Brain Differences in Children With the FTO Obesity Risk Allele. Biological Psychiatry, 2019, 85, S186.	1.3	0
574	Response to Faulkner et al.. Genetics in Medicine, 2021, 23, 243.	2.4	0
575	Newborn screening pilot study in time of pandemic: Duchenne Muscular Dystrophy pilot project experience. Molecular Genetics and Metabolism, 2021, 132, S359.	1.1	0
576	Parental perceptions of genomic sequencing for expanded newborn screening. Molecular Genetics and Metabolism, 2021, 132, S339-S340.	1.1	0

#	Article	IF	Citations
577	Questioning the validity of clinically available breast cancer polygenic risk scores: comparison of two labs reveals discrepancies. Familial Cancer, 2022, 21, 125-127.	1.9	0
578	Familial and Acquired SH2B3 mutations in ALL. Blood, 2012, 120, 1326-1326.	1.4	0
579	Abstract 17301: Functional Characterization of KCNK3 Mutants Associated With Pulmonary Arterial Hypertension Under Physiologically Relevant Heterozygous Conditions. Circulation, 2015, 132, .	1.6	0
580	Genetics of Pulmonary Vascular Disease. , 2016, , 105-121.		0
581	User engagement with web-based genomics education videos and implications for designing scalable patient education materials. AMIA ... Annual Symposium proceedings, 2019, 2019, 923-932.	0.2	0
582	A Novel On-Site Volunteer Community Infection Prevention Team Prevented Outbreaks at a Hurricane Harvey Mega-Shelter. Infection Control and Hospital Epidemiology, 2020, 41, s100-s100.	1.8	0
583	eP123: Design of a phase 2, double-blind, placebo-controlled trial of setmelanotide in patients with genetic variants in the melanocortin-4 receptor pathway. Genetics in Medicine, 2022, 24, S77.	2.4	0
584	PH Roundtable: Genetics and Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 168-175.	0.1	0
585	Genomics of Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 142-149.	0.1	0
586	Discovering the Developmental Basis of Tracheaâ€Esophageal Birth Defects: Evidence for Endosomeâ€opathies. FASEB Journal, 2022, 36, .	0.5	0