Wendy K Chung

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3477328/publications.pdf

Version: 2024-02-01

229	12,225	43973	37111
papers	citations	48 h-index	96 g-index

236	236	236	20476
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	6.0	646
2	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	9.4	624
3	A Novel Channelopathy in Pulmonary Arterial Hypertension. New England Journal of Medicine, 2013, 369, 351-361.	13.9	412
4	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756.	13.7	369
5	Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2013, 62, D13-D21.	1.2	367
6	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
7	Glucocerebrosidase activity in Parkinsonâ€™s disease with and withoutGBAmutations. Brain, 2015, 138, 2648-2658.	3.7	326
8	BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. Lancet Respiratory Medicine,the, 2016, 4, 129-137.	5.2	307
9	Genetics and genomics of pulmonary arterial hypertension. European Respiratory Journal, 2019, 53, 1801899.	3.1	306
10	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	3.8	258
11	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.	2.6	230
12	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.	2.0	229
13	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.	1.1	227
14	Pediatric Cardiomyopathies. Circulation Research, 2017, 121, 855-873.	2.0	207
15	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.	4.3	204
16	The usefulness of whole-exome sequencing in routine clinical practice. Genetics in Medicine, 2014, 16, 922-931.	1.1	196
17	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	1.1	185
18	EIF2AK4 Mutations in Pulmonary Capillary Hemangiomatosis. Chest, 2014, 145, 231-236.	0.4	176

#	Article	IF	Citations
19	Comparison of Parkinson Risk in Ashkenazi Jewish Patients With Gaucher Disease andGBAHeterozygotes. JAMA Neurology, 2014, 71, 752.	4.5	172
20	Opposing Brain Differences in 16p11.2 Deletion and Duplication Carriers. Journal of Neuroscience, 2014, 34, 11199-11211.	1.7	149
21	Implementation of next generation sequencing into pediatric hematology-oncology practice: moving beyond actionable alterations. Genome Medicine, 2016, 8, 133.	3.6	147
22	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	2.6	137
23	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	2.6	134
24	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.	0.7	131
25	CANOES: detecting rare copy number variants from whole exome sequencing data. Nucleic Acids Research, 2014, 42, e97-e97.	6.5	123
26	Complex Genetics and the Etiology of Human Congenital Heart Disease. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a013953-a013953.	2.9	118
27	10-year performance of four models of breast cancer risk: a validation study. Lancet Oncology, The, 2019, 20, 504-517.	5.1	116
28	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	3.6	112
29	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	2.6	110
30	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	1.6	104
31	Precision medicine in diabetes: a Consensus Report from the American Diabetes Association (ADA) and the European Association for the Study of Diabetes (EASD). Diabetologia, 2020, 63, 1671-1693.	2.9	102
32	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	9.4	97
33	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69.	3.6	86
34	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	7.7	83
35	The Congenital Heart Disease Genetic Network Study: Cohort description. PLoS ONE, 2018, 13, e0191319.	1.1	82
36	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.	1.1	81

#	Article	IF	Citations
37	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.	1.1	79
38	Genetic causes of congenital diaphragmatic hernia. Seminars in Fetal and Neonatal Medicine, 2014, 19, 324-330.	1.1	77
39	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	2.6	75
40	Novel Mutations and Decreased Expression of the Epigenetic Regulator TET2 in Pulmonary Arterial Hypertension. Circulation, 2020, 141, 1986-2000.	1.6	75
41	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.	2.2	73
42	Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547.	1.5	68
43	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.	2.6	66
44	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.	1.1	64
45	De novo missense variants in PPP2R5D are associated with intellectual disability, macrocephaly, hypotonia, and autism. Neurogenetics, 2016, 17, 43-49.	0.7	61
46	FTO genotype impacts food intake and corticolimbic activation. American Journal of Clinical Nutrition, 2018, 107, 145-154.	2.2	60
47	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	2.0	60
48	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.	2.6	59
49	Informed consent for return of incidental findings in genomic research. Genetics in Medicine, 2014, 16, 367-373.	1.1	58
50	Mutation inSNAP25as a novel genetic cause of epilepsy and intellectual disability. Rare Diseases (Austin, Tex), 2013, 1, e26314.	1.8	55
51	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.	1.1	55
52	Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314.	0.7	54
53	De novo POGZ mutations are associated with neurodevelopmental disorders and microcephaly. Journal of Physical Education and Sports Management, 2015, 1, a000455.	0.5	51
54	Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824.	5.8	51

#	Article	IF	Citations
55	Age-specific breast cancer risk by body mass index and familial risk: prospective family study cohort (ProF-SC). Breast Cancer Research, 2018, 20, 132.	2.2	51
56	Is there a duty to reinterpret genetic data? The ethical dimensions. Genetics in Medicine, 2020, 22, 633-639.	1.1	51
57	De novo mutations in PURA are associated with hypotonia and developmental delay. Journal of Physical Education and Sports Management, 2015, 1, a000356.	0.5	48
58	Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635.	1.5	48
59	Characterization of a caveolinâ€1 mutation associated with both pulmonary arterial hypertension and congenital generalized lipodystrophy. Traffic, 2016, 17, 1297-1312.	1.3	48
60	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
61	Whole exome sequencing reveals de novo pathogenic variants in KAT6A as a cause of a neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2016, 170, 1791-1798.	0.7	47
62	De novo mutations in CSNK2A1 are associated with neurodevelopmental abnormalities and dysmorphic features. Human Genetics, 2016, 135, 699-705.	1.8	47
63	Frequency of GBA Variants in Autopsyâ€proven Multiple System Atrophy. Movement Disorders Clinical Practice, 2017, 4, 574-581.	0.8	47
64	Clinical and genetic characterization of AP4B1â€associated SPG47. American Journal of Medical Genetics, Part A, 2018, 176, 311-318.	0.7	47
65	De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	1.5	46
66	Identification and characterization of a novel DGAT1 missense mutation associated with congenital diarrhea. Journal of Lipid Research, 2017, 58, 1230-1237.	2.0	44
67	Ethical Considerations Related to Return of Results from Genomic Medicine Projects: The eMERGE Network (Phase III) Experience. Journal of Personalized Medicine, 2018, 8, 2.	1.1	44
68	Robust identification of mosaic variants in congenital heart disease. Human Genetics, 2018, 137, 183-193.	1.8	43
69	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.	3.6	43
70	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.	0.5	42
71	Recent insights into peroxisome biogenesis and associated diseases. Journal of Cell Science, 2020, 133, .	1.2	41
72	Risk-reducing salpingo-oophorectomy, natural menopause, and breast cancer risk: an international prospective cohort of BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2020, 22, 8.	2.2	41

#	Article	IF	Citations
73	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	1.8	40
74	Bitter taste phenotype and body weight predict children's selection of sweet and savory foods at a palatable test-meal. Appetite, 2014, 77, 115-123.	1.8	39
75	Research Participantsâ€™ Preferences for Hypothetical Secondary Results from Genomic Research. Journal of Genetic Counseling, 2017, 26, 841-851.	0.9	39
76	The FTO Gene and Measured Food Intake in 5â€•to 10â€Yearâ€Old Children Without Obesity. Obesity, 2019, 27, 1023-1029.	1.5	39
77	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.	2.6	38
78	Reciprocal white matter alterations due to 16p11.2 chromosomal deletions versus duplications. Human Brain Mapping, 2016, 37, 2833-2848.	1.9	37
79	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.	0.4	37
80	SCARB2 variants and glucocerebrosidase activity in Parkinsonâ€™s disease. Npj Parkinson's Disease, 2016, 2, .	2.5	36
81	Comparison of Clinical, Maternal, and Self Pubertal Assessments: Implications for Health Studies. Pediatrics, 2016, 138, .	1.0	36
82	A human de novo mutation inMYH10phenocopies the loss of function mutation in mice. Rare Diseases (Austin, Tex), 2013, 1, e26144.	1.8	34
83	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	1.1	34
84	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	3.0	34
85	Genotype and defects in microtubule-based motility correlate with clinical severity in KIF1A-associated neurological disorder. Human Genetics and Genomics Advances, 2021, 2, 100026.	1.0	34
86	The impact of hereditary cancer gene panels on clinical care and lessons learned. Journal of Physical Education and Sports Management, 2017, 3, a002154.	0.5	33
87	Correlation of DNA methylation levels in blood and saliva DNA in young girls of the LEGACY Girls study. Epigenetics, 2014, 9, 929-933.	1.3	32
88	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	0.7	32
89	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.	1.4	31
90	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.	0.5	31

#	Article	IF	Citations
91	Mutations in HIVEP2 are associated with developmental delay, intellectual disability, and dysmorphic features. Neurogenetics, 2016, 17, 159-164.	0.7	31
92	Risk-Reducing Oophorectomy and Breast Cancer Risk Across the Spectrum of Familial Risk. Journal of the National Cancer Institute, 2019, 111, 331-334.	3.0	31
93	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	2.6	31
94	Role of Aberrant Spontaneous Neurotransmission in SNAP25-Associated Encephalopathies. Neuron, 2021, 109, 59-72.e5.	3.8	31
95	Generalizability of Polygenic Risk Scores for Breast Cancer Among Women With European, African, and Latinx Ancestry. JAMA Network Open, 2021, 4, e2119084.	2.8	31
96	GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. ELife, 2020, 9, .	2.8	31
97	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.	3.3	30
98	The genetics of isolated congenital heart disease. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 97-106.	0.7	30
99	Auditory Evoked M100 Response Latency is Delayed in Children with 16p11.2 Deletion but not 16p11.2 Duplication. Cerebral Cortex, 2016, 26, 1957-1964.	1.6	29
100	Precision Medicine in Children and Young Adults with Hematologic Malignancies and Blood Disorders: The Columbia University Experience. Frontiers in Pediatrics, 2017, 5, 265.	0.9	29
101	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	1.6	29
102	The Challenge of Genetic Variants of Uncertain Clinical Significance. Annals of Internal Medicine, 2022, 175, 994-1000.	2.0	29
103	Processes and factors involved in decisions regarding return of incidental genomic findings in research. Genetics in Medicine, 2014, 16, 311-317.	1.1	28
104	De novo variants in EBF3 are associated with hypotonia, developmental delay, intellectual disability, and autism. Journal of Physical Education and Sports Management, 2017, 3, a002097.	0.5	28
105	Novel frameshift mutation in Troponin C (TNNC1) associated with hypertrophic cardiomyopathy and sudden death. Cardiology in the Young, 2011, 21, 345-348.	0.4	27
106	De novo truncating variants in the AHDC1 gene encoding the AT-hook DNA-binding motif-containing protein 1 are associated with intellectual disability and developmental delay. Journal of Physical Education and Sports Management, 2015, 1, a000562.	0.5	27
107	Overcoming challenges to meaningful informed consent for whole genome sequencing in pediatric cancer research. Pediatric Blood and Cancer, 2015, 62, 1374-1380.	0.8	27
108	Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829.	1.5	27

#	Article	IF	Citations
109	Alcohol consumption, cigarette smoking, and familial breast cancer risk: findings from the Prospective Family Study Cohort (ProF-SC). Breast Cancer Research, 2019, 21, 128.	2.2	27
110	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.	0.9	27
111	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	2.0	27
112	Impact of Receiving Secondary Results from Genomic Research: A 12â€Month Longitudinal Study. Journal of Genetic Counseling, 2018, 27, 709-722.	0.9	26
113	Participant choices for return of genomic results in the eMERGE Network. Genetics in Medicine, 2020, 22, 1821-1829.	1.1	25
114	United States Pulmonary Hypertension Scientific Registry. Chest, 2021, 159, 311-327.	0.4	25
115	Deep Genetic Connection Between Cancer and Developmental Disorders. Human Mutation, 2016, 37, 1042-1050.	1.1	24
116	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	1.1	23
117	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.	2.1	23
118	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	1.1	23
119	Applying Deep Neural Network Analysis to High-Content Image-Based Assays. SLAS Discovery, 2019, 24, 829-841.	1.4	22
120	Comparative outcomes of right versus left congenital diaphragmatic hernia: A multicenter analysis. Journal of Pediatric Surgery, 2020, 55, 33-38.	0.8	22
121	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
122	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.	1.5	22
123	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.	2.6	22
124	Likely damaging de novo variants in congenital diaphragmatic hernia patients are associated with worse clinical outcomes. Genetics in Medicine, 2020, 22, 2020-2028.	1.1	21
125	Developmental basis of trachea-esophageal birth defects. Developmental Biology, 2021, 477, 85-97.	0.9	21
126	The genetic architecture of pediatric cardiomyopathy. American Journal of Human Genetics, 2022, 109, 282-298.	2.6	21

#	Article	IF	Citations
127	Genetics and Other Omics in Pediatric Pulmonary Arterial Hypertension. Chest, 2020, 157, 1287-1295.	0.4	20
128	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
129	Abnormal auditory and language pathways in children with 16p11.2 deletion. NeuroImage: Clinical, 2015, 9, 50-57.	1.4	19
130	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	1.4	19
131	Neurogenetic disorders across the lifespan: from aberrant development to degeneration. Nature Reviews Neurology, 2022, 18, 117-124.	4.9	19
132	Loss-of-function variants in TIAM1 are associated with developmental delay, intellectual disability, and seizures. American Journal of Human Genetics, 2022, 109, 571-586.	2.6	19
133	Pubertal development in girls by breast cancer family history: the LEGACY girls cohort. Breast Cancer Research, 2017, 19, 69.	2.2	18
134	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.	0.9	18
135	Impact of patient education videos on genetic counseling outcomes after exome sequencing. Patient Education and Counseling, 2020, 103, 127-135.	1.0	18
136	A disorder-related variant (E420K) of a PP2A-regulatory subunit (PPP2R5D) causes constitutively active AKT-mTOR signaling and uncoordinated cell growth. Journal of Biological Chemistry, 2021, 296, 100313.	1.6	18
137	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study. Circulation, 2022, 145, 877-891.	1.6	18
138	Association of Researcher Characteristics with Views on Return of Incidental Findings from Genomic Research. Journal of Genetic Counseling, 2015, 24, 833-841.	0.9	17
139	COVID-19â€™s Impact on Genetics at One Medical Center in New York. Genetics in Medicine, 2020, 22, 1467-1469.	1.1	17
140	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. Genome Medicine, 2020, 12, 42.	3.6	17
141	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130.	1.4	17
142	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.	1.1	17
143	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.	0.4	16
144	Researchersâ€™ views on informed consent for return of secondary results in genomic research. Genetics in Medicine, 2015, 17, 644-650.	1.1	16

#	Article	IF	Citations
145	Examining the Psychosocial Impact of Genetic Testing for Cardiomyopathies. Journal of Genetic Counseling, 2018, 27, 927-934.	0.9	16
146	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	0.7	16
147	A qualitative study of Latinx parents' experiences of clinical exome sequencing. Journal of Genetic Counseling, 2020, 29, 574-586.	0.9	16
148	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.1	16
149	NMIHBA results from hypomorphic PRUNE1 variants that lack short-chain exopolyphosphatase activity. Human Molecular Genetics, 2021, 29, 3516-3531.	1.4	16
150	Genes that drive the pathobiology of pediatric pulmonary arterial hypertension. Pediatric Pulmonology, 2021, 56, 614-620.	1.0	16
151	Detailed Clinical and Psychological Phenotype of the X-linked HNRNPH2-Related Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e551.	0.9	16
152	EPHX1 mutations cause a lipoatrophic diabetes syndrome due to impaired epoxide hydrolysis and increased cellular senescence. ELife, 2021, 10, .	2.8	16
153	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.	0.9	16
154	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	2.6	16
155	Systems Analysis Implicates WAVE2ÂComplex in the Pathogenesis ofÂDevelopmental Left-Sided ObstructiveÂHeart Defects. JACC Basic To Translational Science, 2020, 5, 376-386.	1.9	15
156	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.	0.8	14
157	Does the law require reinterpretation and return of revised genomic results?. Genetics in Medicine, 2021, 23, 833-836.	1.1	14
158	Penetrance of Breast Cancer Susceptibility Genes from the eMERGE III Network. JNCI Cancer Spectrum, 2021, 5, pkab044.	1.4	14
159	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	1.8	14
160	Population-Based Study of Attitudes towardBRCAGenetic Testing among Orthodox Jewish Women. Breast Journal, 2017, 23, 333-337.	0.4	13
161	A newly identified mutation in the PEX26 gene is associated with a milder form of Zellweger spectrum disorder. Journal of Physical Education and Sports Management, 2019, 5, a003483.	0.5	13
162	Cases in Precision Medicine: The Role of Polygenic Risk Scores in Breast Cancer Risk Assessment. Annals of Internal Medicine, 2021, 174, 408-412.	2.0	13

#	Article	IF	Citations
163	Comparing 5-Year and Lifetime Risks of Breast CancerÂusing the Prospective Family Study Cohort. Journal of the National Cancer Institute, 2021, 113, 785-791.	3.0	13
164	Pulmonary arterial hypertension: Specialistsâ€™ knowledge, practices, and attitudes of genetic counseling and genetic testing in the USA. Pulmonary Circulation, 2017, 7, 372-383.	0.8	12
165	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.	0.5	12
166	Imputing cognitive impairment in <scp>SPARK</scp>, a large autism cohort. Autism Research, 2022, 15, 156-170.	2.1	12
167	Do research participants share genomic screening results with family members?. Journal of Genetic Counseling, 2022, 31, 447-458.	0.9	12
168	Recurrent diffuse lung disease due to surfactant protein C deficiency. Respiratory Medicine Case Reports, 2018, 25, 91-95.	0.2	11
169	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.	1.8	11
170	Celiac disease and Down syndrome mortality: a nationwide cohort study. BMC Pediatrics, 2017, 17, 41.	0.7	10
171	VAC14 syndrome in two siblings with retinitis pigmentosa and neurodegeneration with brain iron accumulation. Journal of Physical Education and Sports Management, 2019, 5, a003715.	0.5	10
172	Biallelic variants in AGMO with diminished enzyme activity are associated with a neurodevelopmental disorder. Human Genetics, 2019, 138, 1259-1266.	1.8	10
173	Evaluation of the cost and effectiveness of diverse recruitment methods for a genetic screening study. Genetics in Medicine, 2019, 21, 2371-2380.	1.1	10
174	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	1.1	10
175	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.	2.2	10
176	When to Offer Genetic Testing for Pulmonary Arterial Hypertension. Canadian Journal of Cardiology, 2015, 31, 544-547.	0.8	9
177	Comparison of methods to assess onset of breast development in the LEGACY Girls Study: methodological considerations for studies of breast cancer. Breast Cancer Research, 2018, 20, 33.	2.2	9
178	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.	2.3	9
179	Research on COVID-19 through patient-reported data: a survey for observational studies in the COVID-19 pandemic. Journal of Clinical and Translational Science, 2021, 5, .	0.3	9
180	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.	0.4	9

#	Article	IF	Citations
181	Neurodevelopmental phenotypes in individuals with pathogenic variants in CHAMP1. Journal of Physical Education and Sports Management, 2021, 7, a006092.	0.5	9
182	Whole-Exome Sequencing Reveals CLCNKB Mutations in a Case of Sudden Unexpected Infant Death. Pediatric and Developmental Pathology, 2015, 18, 324-326.	0.5	8
183	Circulating growth factor concentrations and breast cancer risk: a nested case-control study of IGF-1, IGFBP-3, and breast cancer in a family-based cohort. Breast Cancer Research, 2020, 22, 109.	2.2	8
184	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.	1.1	8
185	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.	1.6	8
186	Hypertrophic cardiomyopathy: New approaches and a time to reappraise older approaches. Journal of Thoracic and Cardiovascular Surgery, 2016, 152, 983-988.	0.4	7
187	United States Pulmonary Hypertension Scientific Registry (USPHSR): rationale, design, and clinical implications. Pulmonary Circulation, 2019, 9, 204589401985169.	0.8	7
188	Association of Prepubertal and Adolescent Androgen Concentrations With Timing of Breast Development and Family History of Breast Cancer. JAMA Network Open, 2019, 2, e190083.	2.8	7
189	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	1.0	7
190	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	1.1	7
191	<scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540.	1.0	7
192	Improving Recruitment for a Newborn Screening Pilot Study with Adaptations in Response to the COVID-19 Pandemic. International Journal of Neonatal Screening, 2022, 8, 23.	1.2	7
193	Dilated cardiomyopathy due to a phospholamban duplication. Cardiology in the Young, 2014, 24, 953-954.	0.4	6
194	Human iPSC-Derived Neuronal Cells From CTBP1-Mutated Patients Reveal Altered Expression of Neurodevelopmental Gene Networks. Frontiers in Neuroscience, 2020, 14, 562292.	1.4	6
195	Cross-sectional, quantitative analysis of motor function in females with HNRNPH2-related disorder. Research in Developmental Disabilities, 2021, 119, 104110.	1.2	6
196	Channelopathy Genes in Pulmonary Arterial Hypertension. Biomolecules, 2022, 12, 265.	1.8	6
197	Characterization of phenotypic range in <scp> DYRK1A </scp> haploinsufficiency syndrome using standardized behavioral measures. American Journal of Medical Genetics, Part A, 2022, , .	0.7	6
198	Cancer Genetic Counselor Information Needs for Risk Communication: A Qualitative Evaluation of Interview Transcripts. Journal of Personalized Medicine, 2013, 3, 238-250.	1.1	5

#	Article	IF	Citations
199	Human Subjects Protection: An Event Monitoring Committee for Research Studies of Girls From Breast Cancer Families. Journal of Adolescent Health, 2014, 55, 352-357.	1.2	5
200	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.	1.0	5
201	Genetic Variant Reinterpretation: Economic and Population Health Management Challenges. Population Health Management, 2021, 24, 310-313.	0.8	5
202	Neither cardiac mitochondrial DNA variation nor copy number contribute to congenital heart disease risk. American Journal of Human Genetics, 2022, 109, 961-966.	2.6	5
203	Choices, attitudes, and experiences of genetic screening in Latino/a and Ashkenazi Jewish individuals. Journal of Community Genetics, 2020, 11, 391-403.	0.5	4
204	Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes. Genome Medicine, 2021, 13, 147.	3.6	4
205	Extracorporeal membrane oxygenation (ECMO) and its complications in newborns with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2022, , .	0.8	4
206	Weight is More Informative than Body Mass Index for Predicting Postmenopausal Breast Cancer Risk: Prospective Family Study Cohort (ProF-SC). Cancer Prevention Research, 2022, 15, 185-191.	0.7	4
207	Heterozygous variants in PRPF8 are associated with neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2022, 188, 2750-2759.	0.7	4
208	Mutations in BMPR2 are not present in patients with pulmonary hypertension associated with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2017, 52, 1747-1750.	0.8	3
209	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. Human Mutation, 2018, 39, 870-881.	1.1	3
210	Clinical and genomic characterization of 8p cytogenomic disorders. Genetics in Medicine, 2021, 23, 2342-2351.	1.1	3
211	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.	1.1	3
212	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
213	ACSS2 gene variant associated with cleft lip and palate in two independent Hispanic populations. Laryngoscope, 2017, 127, E336-E339.	1.1	2
214	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.	1.6	2
215	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.	1.0	2
216	Considerations When Using Breast Cancer Risk Models for Women with Negative BRCA1/BRCA2 Mutation Results. Journal of the National Cancer Institute, 2020, 112, 418-422.	3.0	1

#	Article	IF	Citations
217	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.	1.1	1
218	Developing effective and efficient genomic educational tools for our diverse population. Annals of Translational Medicine, 2019, 7, S304-S304.	0.7	1
219	Recreational Physical Activity and Outcomes After Breast Cancer in Women at High Familial Risk. JNCI Cancer Spectrum, 2021, 5, pkab090.	1.4	1
220	Maternal and prenatal factors and age at thelarche in the LEGACY Girls Study cohort: implications for breast cancer risk. International Journal of Epidemiology, 2023, 52, 272-283.	0.9	1
221	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, , .	1.7	1
222	Partial Duplication and Poly(A) Insertion in KCNQ1 Not Detected by Next-Generation Sequencing in Jervell and Langeâ€“Nielsen Syndrome. Circulation: Arrhythmia and Electrophysiology, 2016, 9, .	2.1	0
223	Genetics and Hypertrophic Cardiomyopathy. Current Pediatrics Reports, 2016, 4, 35-44.	1.7	0
224	Pediatric genetics: rare is common. Journal of Physical Education and Sports Management, 2020, 6, a005587.	0.5	0
225	Response to Faulkner et al.. Genetics in Medicine, 2021, 23, 243.	1.1	0
226	Questioning the validity of clinically available breast cancer polygenic risk scores: comparison of two labs reveals discrepancies. Familial Cancer, 2022, 21, 125-127.	0.9	0
227	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
228	OUP accepted manuscript. International Journal of Epidemiology, 2022, , .	0.9	0
229	Genomics of Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 142-149.	0.1	0