Wendy K Chung

List of Publications by Year
in descending order

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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	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
2	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
3	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
4	Imputing cognitive impairment in <scp>SPARK</scp>, a large autism cohort. Autism Research, 2022, 15, 156-170.	2.1	12
5	Do research participants share genomic screening results with family members?. Journal of Genetic Counseling, 2022, 31, 447-458.	0.9	12
6	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
7	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	1.1	7
8	The genetic architecture of pediatric cardiomyopathy. American Journal of Human Genetics, 2022, 109, 282-298.	2.6	21
9	Extracorporeal membrane oxygenation (ECMO) and its complications in newborns with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2022, , .	0.8	4
10	Neurogenetic disorders across the lifespan: from aberrant development to degeneration. Nature Reviews Neurology, 2022, 18, 117-124.	4.9	19
11	OUP accepted manuscript. International Journal of Epidemiology, 2022, , .	0.9	0
12	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
13	Channelopathy Genes in Pulmonary Arterial Hypertension. Biomolecules, 2022, 12, 265.	1.8	6
14	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study. Circulation, 2022, 145, 877-891.	1.6	18
15	<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
16	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
17	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
18	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

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19	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
20	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
21	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
22	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
23	The Challenge of Genetic Variants of Uncertain Clinical Significance. Annals of Internal Medicine, 2022, 175, 994-1000.	2.0	29
24	Heterozygous variants in PRPF8 are associated with neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2022, 188, 2750-2759.	0.7	4
25	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
26	Response to Faulkner et al.. Genetics in Medicine, 2021, 23, 243.	1.1	0
27	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	1.6	29
28	Role of Aberrant Spontaneous Neurotransmission in SNAP25-Associated Encephalopathies. Neuron, 2021, 109, 59-72.e5.	3.8	31
29	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	1.0	7
30	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
31	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130.	1.4	17
32	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	3.0	34
33	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
34	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
35	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
36	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

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37	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
38	NMIHBA results from hypomorphic PRUNE1 variants that lack short-chain exopolyphosphatase activity. Human Molecular Genetics, 2021, 29, 3516-3531.	1.4	16
39	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
40	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
41	Genetic Variant Reinterpretation: Economic and Population Health Management Challenges. Population Health Management, 2021, 24, 310-313.	0.8	5
42	United States Pulmonary Hypertension Scientific Registry. Chest, 2021, 159, 311-327.	0.4	25
43	Genes that drive the pathobiology of pediatric pulmonary arterial hypertension. Pediatric Pulmonology, 2021, 56, 614-620.	1.0	16
44	Detailed Clinical and Psychological Phenotype of the X-linked HNRNPH2-Related Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e551.	0.9	16
45	Does the law require reinterpretation and return of revised genomic results?. Genetics in Medicine, 2021, 23, 833-836.	1.1	14
46	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
47	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	2.0	27
48	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
49	Penetrance of Breast Cancer Susceptibility Genes from the eMERGE III Network. JNCI Cancer Spectrum, 2021, 5, pkab044.	1.4	14
50	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
51	Neurodevelopmental phenotypes in individuals with pathogenic variants in CHAMP1. Journal of Physical Education and Sports Management, 2021, 7, a006092.	0.5	9
52	Clinical and genomic characterization of 8p cytogenomic disorders. Genetics in Medicine, 2021, 23, 2342-2351.	1.1	3
53	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
54	EPHX1 mutations cause a lipoatrophic diabetes syndrome due to impaired epoxide hydrolysis and increased cellular senescence. ELife, 2021, 10, .	2.8	16

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55	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
56	Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes. Genome Medicine, 2021, 13, 147.	3.6	4
57	Developmental basis of trachea-esophageal birth defects. Developmental Biology, 2021, 477, 85-97.	0.9	21
58	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
59	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
60	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
61	Cross-sectional, quantitative analysis of motor function in females with HNRNPH2-related disorder. Research in Developmental Disabilities, 2021, 119, 104110.	1.2	6
62	Recreational Physical Activity and Outcomes After Breast Cancer in Women at High Familial Risk. JNCI Cancer Spectrum, 2021, 5, pkab090.	1.4	1
63	Genomics of Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 142-149.	0.1	0
64	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
65	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
66	Is there a duty to reinterpret genetic data? The ethical dimensions. Genetics in Medicine, 2020, 22, 633-639.	1.1	51
67	Comparative outcomes of right versus left congenital diaphragmatic hernia: A multicenter analysis. Journal of Pediatric Surgery, 2020, 55, 33-38.	0.8	22
68	Impact of patient education videos on genetic counseling outcomes after exome sequencing. Patient Education and Counseling, 2020, 103, 127-135.	1.0	18
69	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
70	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
71	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756.	13.7	369
72	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

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73	Pediatric genetics: rare is common. Journal of Physical Education and Sports Management, 2020, 6, a005587.	0.5	0
74	Participant choices for return of genomic results in the eMERGE Network. Genetics in Medicine, 2020, 22, 1821-1829.	1.1	25
75	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
76	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
77	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
78	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
79	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	2.6	110
80	Systems Analysis Implicates WAVE2ÂComplex in the Pathogenesis ofÂDevelopmental Left-Sided ObstructiveÂHeart Defects. JACC Basic To Translational Science, 2020, 5, 376-386.	1.9	15
81	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
82	A qualitative study of Latinx parents' experiences of clinical exome sequencing. Journal of Genetic Counseling, 2020, 29, 574-586.	0.9	16
83	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
84	Recent insights into peroxisome biogenesis and associated diseases. Journal of Cell Science, 2020, 133, .	1.2	41
85	COVID-19â€™s Impact on Genetics at One Medical Center in New York. Genetics in Medicine, 2020, 22, 1467-1469.	1.1	17
86	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
87	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
88	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	1.1	10
89	Novel Mutations and Decreased Expression of the Epigenetic Regulator TET2 in Pulmonary Arterial Hypertension. Circulation, 2020, 141, 1986-2000.	1.6	75
90	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	9.4	97

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91	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
92	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
93	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq1 1 0.784314 rgBT /Overlock	1.1	16
94	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
95	Genetics and Other Omics in Pediatric Pulmonary Arterial Hypertension. Chest, 2020, 157, 1287-1295.	0.4	20
96	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. Genome Medicine, 2020, 12, 42.	3.6	17
97	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	2.6	31
98	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	7.7	83
99	GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. ELife, 2020, 9, .	2.8	31
100	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
101	Applying Deep Neural Network Analysis to High-Content Image-Based Assays. SLAS Discovery, 2019, 24, 829-841.	1.4	22
102	Biallelic variants in AGMO with diminished enzyme activity are associated with a neurodevelopmental disorder. Human Genetics, 2019, 138, 1259-1266.	1.8	10
103	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
104	The FTO Gene and Measured Food Intake in 5â€•to 10â€Yearâ€Old Children Without Obesity. Obesity, 2019, 27, 1023-1029.	1.5	39
105	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
106	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
107	United States Pulmonary Hypertension Scientific Registry (USPHSR): rationale, design, and clinical implications. Pulmonary Circulation, 2019, 9, 204589401985169.	0.8	7
108	A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143.	0.7	16

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109	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
110	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
111	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
112	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	1.4	19
113	10-year performance of four models of breast cancer risk: a validation study. Lancet Oncology, The, 2019, 20, 504-517.	5.1	116
114	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69.	3.6	86
115	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
116	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
117	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
118	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
119	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
120	Genetics and genomics of pulmonary arterial hypertension. European Respiratory Journal, 2019, 53, 1801899.	3.1	306
121	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
122	Developing effective and efficient genomic educational tools for our diverse population. Annals of Translational Medicine, 2019, 7, S304-S304.	0.7	1
123	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
124	FTO genotype impacts food intake and corticolimbic activation. American Journal of Clinical Nutrition, 2018, 107, 145-154.	2.2	60
125	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	1.6	104
126	Robust identification of mosaic variants in congenital heart disease. Human Genetics, 2018, 137, 183-193.	1.8	43

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127	Examining the Psychosocial Impact of Genetic Testing for Cardiomyopathies. Journal of Genetic Counseling, 2018, 27, 927-934.	0.9	16
128	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
129	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
130	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
131	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
132	Impact of Receiving Secondary Results from Genomic Research: A 12â€Month Longitudinal Study. Journal of Genetic Counseling, 2018, 27, 709-722.	0.9	26
133	Clinical and genetic characterization of AP4B1â€associated SPG47. American Journal of Medical Genetics, Part A, 2018, 176, 311-318.	0.7	47
134	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
135	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
136	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	1.1	23
137	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	3.6	112
138	Recurrent diffuse lung disease due to surfactant protein C deficiency. Respiratory Medicine Case Reports, 2018, 25, 91-95.	0.2	11
139	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
140	The Congenital Heart Disease Genetic Network Study: Cohort description. PLoS ONE, 2018, 13, e0191319.	1.1	82
141	Celiac disease and Down syndrome mortality: a nationwide cohort study. BMC Pediatrics, 2017, 17, 41.	0.7	10
142	De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	1.5	46
143	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
144	Research Participantsâ€™ Preferences for Hypothetical Secondary Results from Genomic Research. Journal of Genetic Counseling, 2017, 26, 841-851.	0.9	39

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145	Population-Based Study of Attitudes towardBRCAGenetic Testing among Orthodox Jewish Women. Breast Journal, 2017, 23, 333-337.	0.4	13
146	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
147	Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829.	1.5	27
148	ACSS2 gene variant associated with cleft lip and palate in two independent Hispanic populations. Laryngoscope, 2017, 127, E336-E339.	1.1	2
149	Identification and characterization of a novel DGAT1 missense mutation associated with congenital diarrhea. Journal of Lipid Research, 2017, 58, 1230-1237.	2.0	44
150	Frequency of GBA Variants in Autopsyâ€proven Multiple System Atrophy. Movement Disorders Clinical Practice, 2017, 4, 574-581.	0.8	47
151	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
152	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	9.4	624
153	Pediatric Cardiomyopathies. Circulation Research, 2017, 121, 855-873.	2.0	207
154	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
155	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
156	Pubertal development in girls by breast cancer family history: the LEGACY girls cohort. Breast Cancer Research, 2017, 19, 69.	2.2	18
157	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
158	Hypertrophic cardiomyopathy: New approaches and a time to reappraise older approaches. Journal of Thoracic and Cardiovascular Surgery, 2016, 152, 983-988.	0.4	7
159	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
160	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
161	Implementation of next generation sequencing into pediatric hematology-oncology practice: moving beyond actionable alterations. Genome Medicine, 2016, 8, 133.	3.6	147
162	Mutations in HIVEP2 are associated with developmental delay, intellectual disability, and dysmorphic features. Neurogenetics, 2016, 17, 159-164.	0.7	31

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163	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
164	De novo mutations in CSNK2A1 are associated with neurodevelopmental abnormalities and dysmorphic features. Human Genetics, 2016, 135, 699-705.	1.8	47
165	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
166	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	2.6	137
167	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
168	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	1.8	40
169	Characterization of a caveolinâ€1 mutation associated with both pulmonary arterial hypertension and congenital generalized lipodystrophy. Traffic, 2016, 17, 1297-1312.	1.3	48
170	Deep Genetic Connection Between Cancer and Developmental Disorders. Human Mutation, 2016, 37, 1042-1050.	1.1	24
171	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
172	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
173	Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824.	5.8	51
174	SCARB2 variants and glucocerebrosidase activity in Parkinsonâ€™s disease. Npj Parkinson's Disease, 2016, 2, .	2.5	36
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