Wendy Chung

List of Publications by Year
in descending order

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		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	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
2	Neurodevelopmental phenotypes associated with pathogenic variants in SLC6A1. Journal of Medical Genetics, 2022, 59, 536-543.	3.2	18
3	Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders. Journal of Medical Genetics, 2022, 59, 669-677.	3.2	5
4	Post-translational formation of hypusine in eIF5A: implications in human neurodevelopment. Amino Acids, 2022, 54, 485-499.	2.7	19
5	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
6	Neurodevelopmental profile of HIVEP2â€related disorder. Developmental Medicine and Child Neurology, 2022, 64, 654-661.	2.1	3
7	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
8	Genetics Dictating Therapeutic Decisions in Pediatric Pulmonary Hypertension? A Case Report Suggesting We Are Getting Closer.. Pulmonary Circulation, 2022, 12, e12033.	1.7	5
9	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
10	The genetic architecture of pediatric cardiomyopathy. American Journal of Human Genetics, 2022, 109, 282-298.	6.2	21
11	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
12	Extracorporeal membrane oxygenation (ECMO) and its complications in newborns with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2022, , .	1.6	4
13	Genotype-Phenotype Comparison in POGZ-Related Neurodevelopmental Disorders by Using Clinical Scoring. Genes, 2022, 13, 154.	2.4	6
14	Neurogenetic disorders across the lifespan: from aberrant development to degeneration. Nature Reviews Neurology, 2022, 18, 117-124.	10.1	19
15	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
16	Channelopathy Genes in Pulmonary Arterial Hypertension. Biomolecules, 2022, 12, 265.	4.0	6
17	The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network. Genetics in Medicine, 2022, 24, 1130-1138.	2.4	12
18	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study. Circulation, 2022, 145, 877-891.	1.6	18

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19	OP035: Rapid Whole Genome Sequencing (rWGS) in the cardiac NICU. Genetics in Medicine, 2022, 24, S362-S363.	2.4	1
20	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
21	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
22	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
23	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
24	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
25	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
26	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
27	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835.	7.1	25
28	Discovering the Developmental Basis of Tracheaâ€Esophageal Birth Defects: Evidence for Endosomeâ€opathies. FASEB Journal, 2022, 36, .	0.5	0
29	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
30	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
31	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
32	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
33	Response to Faulkner et al.. Genetics in Medicine, 2021, 23, 243.	2.4	0
34	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
35	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
36	Common Childhood Viruses and Pubertal Timing: The LEGACY Girls Study. American Journal of Epidemiology, 2021, 190, 766-778.	3.4	3

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37	Role of Aberrant Spontaneous Neurotransmission in SNAP25-Associated Encephalopathies. Neuron, 2021, 109, 59-72.e5.	8.1	31
38	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	2.0	7
39	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130.	2.8	17
40	Prepubertal Internalizing Symptoms and Timing of Puberty Onset in Girls. American Journal of Epidemiology, 2021, 190, 431-438.	3.4	14
41	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	6.1	34
42	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
43	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
44	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
45	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
46	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
47	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
48	Reply to â€œ<scp>PPP2R5D</scp> Genetic Mutations and Early Onset Parkinsonismâ€• Annals of Neurology, 2021, 89, 195-196.	5.3	1
49	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
50	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
51	Genetic Variant Reinterpretation: Economic and Population Health Management Challenges. Population Health Management, 2021, 24, 310-313.	1.7	5
52	United States Pulmonary Hypertension Scientific Registry. Chest, 2021, 159, 311-327.	0.8	25
53	Genes that drive the pathobiology of pediatric pulmonary arterial hypertension. Pediatric Pulmonology, 2021, 56, 614-620.	2.0	16
54	Detailed Clinical and Psychological Phenotype of the X-linked HNRNPH2-Related Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e551.	1.9	16

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55	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
56	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
57	MVP predicts theÂpathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	12.8	85
58	An electronic health record (EHR) log analysis shows limited clinician engagement with unsolicited genetic test results. JAMIA Open, 2021, 4, ooab014.	2.0	5
59	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
60	Does the law require reinterpretation and return of revised genomic results?. Genetics in Medicine, 2021, 23, 833-836.	2.4	14
61	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
62	The psychiatric phenotypes of 1q21 distal deletion and duplication. Translational Psychiatry, 2021, 11, 105.	4.8	6
63	Early Pandemic Experiences of Autistic Adults: Predictors of Psychological Distress. Autism Research, 2021, 14, 1209-1219.	3.8	48
64	Response to Li and Hopper. American Journal of Human Genetics, 2021, 108, 527-529.	6.2	5
65	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
66	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
67	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
68	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
69	Newborn screening pilot study in time of pandemic: Duchenne Muscular Dystrophy pilot project experience. Molecular Genetics and Metabolism, 2021, 132, S359.	1.1	0
70	Reproductive decisionâ€making in families containing multiple individuals with epilepsy. Epilepsia, 2021, 62, 1220-1230.	5.1	5
71	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	4.5	27
72	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

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73	Parental perceptions of genomic sequencing for expanded newborn screening. Molecular Genetics and Metabolism, 2021, 132, S339-S340.	1.1	0
74	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
75	Penetrance of Breast Cancer Susceptibility Genes from the eMERGE III Network. JNCI Cancer Spectrum, 2021, 5, pkab044.	2.9	14
76	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
77	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
78	Genomic medicine implementation protocols in the PhenX Toolkit: tools for standardized data collection. Genetics in Medicine, 2021, 23, 1783-1788.	2.4	2
79	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
80	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
81	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
82	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
83	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
84	Neurodevelopmental phenotypes in individuals with pathogenic variants in CHAMP1. Journal of Physical Education and Sports Management, 2021, 7, a006092.	1.2	9
85	16p11.2 deletion syndrome. Current Opinion in Genetics and Development, 2021, 68, 49-56.	3.3	39
86	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
87	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
88	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
89	Clinical and genomic characterization of 8p cytogenomic disorders. Genetics in Medicine, 2021, 23, 2342-2351.	2.4	3
90	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	2.4	3

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91	Case Report: Esophageal Bronchus in a Neonate, With Image, Histological, and Molecular Analysis. Frontiers in Pediatrics, 2021, 9, 707822.	1.9	3
92	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
93	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
94	Harnessing rare variants in neuropsychiatric and neurodevelopment disordersâ€"a Keystone Symposia report. Annals of the New York Academy of Sciences, 2021, , .	3.8	2
95	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
96	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
97	Gene expression atlas of energy balance brain regions. JCI Insight, 2021, 6, .	5.0	6
98	Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes. Genome Medicine, 2021, 13, 147.	8.2	4
99	A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. New England Journal of Medicine, 2021, 385, 1292-1301.	27.0	23
100	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
101	Developmental basis of trachea-esophageal birth defects. Developmental Biology, 2021, 477, 85-97.	2.0	21
102	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
103	Implementation of Population-Based Newborn Screening Reveals Low Incidence of Spinal Muscular Atrophy. Obstetrical and Gynecological Survey, 2021, 76, 17-19.	0.4	1
104	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
105	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
106	Biodistribution of onasemnogene abeparvovec DNA, mRNA and SMN protein in human tissue. Nature Medicine, 2021, 27, 1701-1711.	30.7	49
107	Reimbursement for genetic variant reinterpretation: five questions payers should ask. American Journal of Managed Care, 2021, 27, e336-e338.	1.1	3
108	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

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109	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
110	Cross-sectional, quantitative analysis of motor function in females with HNRNPH2-related disorder. Research in Developmental Disabilities, 2021, 119, 104110.	2.2	6
111	Recreational Physical Activity and Outcomes After Breast Cancer in Women at High Familial Risk. JNCI Cancer Spectrum, 2021, 5, pkab090.	2.9	1
112	PH Roundtable: Genetics and Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 168-175.	0.1	0
113	Genomics of Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 142-149.	0.1	0
114	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
115	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
116	Is there a duty to reinterpret genetic data? The ethical dimensions. Genetics in Medicine, 2020, 22, 633-639.	2.4	51
117	The influence of genetics in congenital diaphragmatic hernia. Seminars in Perinatology, 2020, 44, 151169.	2.5	39
118	Comparative outcomes of right versus left congenital diaphragmatic hernia: A multicenter analysis. Journal of Pediatric Surgery, 2020, 55, 33-38.	1.6	22
119	Psychotic symptoms in 16p11.2 copyâ€number variant carriers. Autism Research, 2020, 13, 187-198.	3.8	11
120	Impact of patient education videos on genetic counseling outcomes after exome sequencing. Patient Education and Counseling, 2020, 103, 127-135.	2.2	18
121	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
122	Genetic Basis of Human Congenital Heart Disease. Cold Spring Harbor Perspectives in Biology, 2020, 12, a036749.	5.5	34
123	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
124	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756.	27.8	369
125	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
126	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

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127	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
128	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
129	Common germline-somatic variant interactions in advanced urothelial cancer. Nature Communications, 2020, 11, 6195.	12.8	21
130	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
131	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
132	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444.	6.2	124
133	Participant choices for return of genomic results in the eMERGE Network. Genetics in Medicine, 2020, 22, 1821-1829.	2.4	25
134	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
135	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
136	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
137	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
138	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
139	Genetics and Genomics of Pediatric Pulmonary Arterial Hypertension. Genes, 2020, 11, 1213.	2.4	24
140	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
141	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
142	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
143	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	6.2	110
144	Implementation of population-based newborn screening reveals low incidence of spinal muscular atrophy. Genetics in Medicine, 2020, 22, 1296-1302.	2.4	57

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145	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
146	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
147	A qualitative study of Latinx parents' experiences of clinical exome sequencing. Journal of Genetic Counseling, 2020, 29, 574-586.	1.6	16
148	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
149	Returning Results in the Genomic Era: Initial Experiences of the eMERGE Network. Journal of Personalized Medicine, 2020, 10, 30.	2.5	39
150	Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477.	2.4	61
151	COVID-19â€™s Impact on Genetics at One Medical Center in New York. Genetics in Medicine, 2020, 22, 1467-1469.	2.4	17
152	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
153	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
154	Novel Mutations and Decreased Expression of the Epigenetic Regulator TET2 in Pulmonary Arterial Hypertension. Circulation, 2020, 141, 1986-2000.	1.6	75
155	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	21.4	97
156	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
157	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
158	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
159	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq1 1 0.784314 rgBT /Overlo	2.5	16
160	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
161	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
162	Psychiatric and Medical Profiles of Autistic Adults in the SPARK Cohort. Journal of Autism and Developmental Disorders, 2020, 50, 3679-3698.	2.7	33

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163	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
164	Genetics and Other Omics in Pediatric Pulmonary Arterial Hypertension. Chest, 2020, 157, 1287-1295.	0.8	20
165	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
166	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	6.2	31
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