Wendy Chung

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

561 papers	30,793 citations	81 h-index	159 g-index
621 ext. papers	39,617 ext. citations	7.4 avg, IF	6.72 L-index

#	Paper	IF	Citations
561	Cancer Risks Associated With and Pathogenic Variants.. *Journal of Clinical Oncology*, **2022**, JCO2102112	2.2	7
560	Neurogenetic disorders across the lifespan: from aberrant development to degeneration.. *Nature Reviews Neurology*, **2022**,	15	4
559	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity.. *Circulation Genomic and Precision Medicine*, **2022**, CIRCGEN121003500	5.2	0
558	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability.. *Human Genetics and Genomics Advances*, **2022**, 3, 100074	0.8	3
557	Genetics dictating therapeutic decisions in pediatric pulmonary hypertension? A case report suggesting we are getting closer.. *Pulmonary Circulation*, **2022**, 12, e12033	2.7	0
556	The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network.. *Genetics in Medicine*, **2022**,	8.1	2
555	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. *American Journal of Human Genetics*, **2022**, 109, 601-617	11	0
554	Neither cardiac mitochondrial DNA variation or copy number contribute to congenital heart disease risk.. *American Journal of Human Genetics*, **2022**,	11	1
553	Generation of three induced pluripotent stem cells lines from patients with esophageal atresia/tracheoesophageal fistula type C.. *Stem Cell Research*, **2022**, 60, 102711	1.6	0
552	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	0.8	0
551	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B.. *Clinical Epigenetics*, **2022**, 14, 52	7.7	1
550	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases.. *JAMA Oncology*, **2022**,	13.4	2
549	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	11	0
548	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,	11.5	1
547	Cross-sectional, quantitative analysis of motor function in females with HNRNPH2-related disorder. *Research in Developmental Disabilities*, **2021**, 119, 104110	2.7	0
546	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	11	3
545	Biodistribution of onasemnogene abeparvovec DNA, mRNA and SMN protein in human tissue. *Nature Medicine*, **2021**, 27, 1701-1711	50.5	8

544	Reimbursement for genetic variant reinterpretation: five questions payers should ask. *American Journal of Managed Care*, **2021**, 27, e336-e338	2.1	0
543	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	2.3	0
542	Response to Faulkner et al. *Genetics in Medicine*, **2021**, 23, 243	8.1	
541	A novel homozygous variant in results in a neurodevelopmental disorder and disrupts TRAPP complex function. *Journal of Medical Genetics*, **2021**, 58, 592-601	5.8	3
540	Response to Li and Hopper. *American Journal of Human Genetics*, **2021**, 108, 527-529	11	1
539	Availability of Services and Caregiver Burden: Supporting Individuals With Neurogenetic Conditions During the COVID-19 Pandemic. *Journal of Child Neurology*, **2021**, 36, 760-767	2.5	3
538	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	8.1	13
537	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	1.6	1
536	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	1.5	0
535	Reproductive decision-making in families containing multiple individuals with epilepsy. *Epilepsia*, **2021**, 62, 1220-1230	6.4	1
534	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. *Circulation Research* , **2021**, 128, 1156-1169	15.7	2
533	Genotype and defects in microtubule-based motility correlate with clinical severity in -associated neurological disorder. *Human Genetics and Genomics Advances*, **2021**, 2,	0.8	9
532	Genetic Causes of Cardiomyopathy in Children: First Results From the Pediatric Cardiomyopathy Genes Study. *Journal of the American Heart Association*, **2021**, 10, e017731	6	11
531	Penetrance of Breast Cancer Susceptibility Genes From the eMERGE III Network. *JNCI Cancer Spectrum*, **2021**, 5, pkab044	4.6	4
530	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	8.1	33
529	Practice Patterns After Return of Rare Variants Associated With Cardiomyopathy in the Electronic Medical Records and Genomics Network. *Circulation: Heart Failure*, **2021**, 14, e008155	7.6	
528	Genomic medicine implementation protocols in the PhenX Toolkit: tools for standardized data collection. *Genetics in Medicine*, **2021**, 23, 1783-1788	8.1	1
527	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	14.4	11

526	Questioning the validity of clinically available breast cancer polygenic risk scores: comparison of two labs reveals discrepancies. *Familial Cancer*, **2021**, 1	3	
525	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	8.1	6
524	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	8.1	81
523	Neurodevelopmental phenotypes associated with pathogenic variants in. *Journal of Medical Genetics*, **2021**,	5.8	2
522	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. *Genetics in Medicine*, **2021**, 23, 1624-1635	8.1	0
521	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	11	2
520	Neurodevelopmental phenotypes in individuals with pathogenic variants in. *Journal of Physical Education and Sports Management*, **2021**, 7,	2.8	1
519	16p11.2 deletion syndrome. *Current Opinion in Genetics and Development*, **2021**, 68, 49-56	4.9	8
518	The predictive ability of the 313 variant-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	8.1	2
517	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	2.3	2
516	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	11	3
515	Delineating the genotypic and phenotypic spectrum of -related neurodevelopmental disorders. *Journal of Medical Genetics*, **2021**,	5.8	1
514	Clinical and genomic characterization of 8p cytogenomic disorders. *Genetics in Medicine*, **2021**, 23, 2342-2351	8.1	0
513	Neptune: an environment for the delivery of genomic medicine. *Genetics in Medicine*, **2021**, 23, 1838-1845	8.1	1
512	Case Report: Esophageal Bronchus in a Neonate, With Image, Histological, and Molecular Analysis. *Frontiers in Pediatrics*, **2021**, 9, 707822	3.4	
511	Common Childhood Viruses and Pubertal Timing: The LEGACY Girls Study. *American Journal of Epidemiology*, **2021**, 190, 766-778	3.8	1
510	Role of Aberrant Spontaneous Neurotransmission in SNAP25-Associated Encephalopathies. *Neuron* , **2021**, 109, 59-72.e5	13.9	7
509	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. *Clinical Genetics*, **2021**, 99, 313-317	4	4

508	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. *European Journal of Human Genetics*, **2021**, 29, 122-130	5.3	6
507	Prepubertal Internalizing Symptoms and Timing of Puberty Onset in Girls. *American Journal of Epidemiology*, **2021**, 190, 431-438	3.8	2
506	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. *JAMA Cardiology*, **2021**, 6, 457-462	16.2	12
505	Cases in Precision Medicine: The Role of Polygenic Risk Scores in Breast Cancer Risk Assessment. *Annals of Internal Medicine*, **2021**, 174, 408-412	8	2
504	Weight-loss response to naltrexone/bupropion is modulated by the Taq1A genetic variant near DRD2 (rs1800497): A pilot study. *Diabetes, Obesity and Metabolism*, **2021**, 23, 850-853	6.7	3
503	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. *Genetics in Medicine*, **2021**, 23, 653-660	8.1	5
502	Comparing 5-Year and Lifetime Risks of Breast Cancer using the Prospective Family Study Cohort. *Journal of the National Cancer Institute*, **2021**, 113, 785-791	9.7	5
501	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	4	2
500	Returning negative results from large-scale genomic screening: Experiences from the eMERGE III network. *American Journal of Medical Genetics, Part A*, **2021**, 185, 508-516	2.5	2
499	Reply to "PPP2R5D Genetic Mutations and Early Onset Parkinsonism". *Annals of Neurology*, **2021**, 89, 195-196	9.4	0
498	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. *Genetics in Medicine*, **2021**, 23, 352-362	8.1	5
497	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	8.1	4
496	Genetic Variant Reinterpretation: Economic and Population Health Management Challenges. *Population Health Management*, **2021**, 24, 310-313	1.8	2
495	United States Pulmonary Hypertension Scientific Registry: Baseline Characteristics. *Chest*, **2021**, 159, 311-327	5.3	7
494	Genes that drive the pathobiology of pediatric pulmonary arterial hypertension. *Pediatric Pulmonology*, **2021**, 56, 614-620	3.5	6
493	Detailed Clinical and Psychological Phenotype of the X-linked -Related Neurodevelopmental Disorder. *Neurology: Genetics*, **2021**, 7, e551	3.8	6
492	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	11.9	21
491	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	4.6	41

490	MVP predicts the pathogenicity of missense variants by deep learning. *Nature Communications*, **2021**, 12, 510	17.4	23
489	An electronic health record (EHR) log analysis shows limited clinician engagement with unsolicited genetic test results. *JAMIA Open*, **2021**, 4, ooab014	2.9	2
488	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	5.3	4
487	Does the law require reinterpretation and return of revised genomic results?. *Genetics in Medicine*, **2021**, 23, 833-836	8.1	4
486	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. *Nature Communications*, **2021**, 12, 1078	17.4	4
485	The psychiatric phenotypes of 1q21 distal deletion and duplication. *Translational Psychiatry*, **2021**, 11, 105	8.6	4
484	Early Pandemic Experiences of Autistic Adults: Predictors of Psychological Distress. *Autism Research*, **2021**, 14, 1209-1219	5.1	23
483	Post-translational formation of hypusine in eIF5A: implications in human neurodevelopment. *Amino Acids*, **2021**, 1	3.5	4
482	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	5.2	
481	Generalizability of Polygenic Risk Scores for Breast Cancer Among Women With European, African, and Latinx Ancestry. *JAMA Network Open*, **2021**, 4, e2119084	10.4	5
480	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. *Genetics in Medicine*, **2021**, 23, 2415-2425	8.1	0
479	Neuropathological Findings in a Case of Parkinsonism and Developmental Delay Associated with a Monoallelic Variant in PLXNA1. *Movement Disorders*, **2021**, 36, 2681-2687	7	1
478	Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes. *Genome Medicine*, **2021**, 13, 147	14.4	0
477	A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. *New England Journal of Medicine*, **2021**, 385, 1292-1301	59.2	6
476	Biallelic variants of cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. *Journal of Medical Genetics*, **2021**,	5.8	3
475	Bi-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**,	2.5	2
474	Developmental basis of trachea-esophageal birth defects. *Developmental Biology*, **2021**, 477, 85-97	3.1	1
473	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	11	6

472	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	2.5	5
471	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study.. *Circulation*, **2021**,	16.7	2
470	Recreational Physical Activity and Outcomes After Breast Cancer in Women at High Familial Risk.. *JNCI Cancer Spectrum*, **2021**, 5, pkab090	4.6	0
469	Genomics of Pulmonary Hypertension. *Advances in Pulmonary Hypertension*, **2021**, 20, 142-149	0.5	
468	Impact of Coronavirus Disease 2019 (COVID-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	6	24
467	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. *American Journal of Human Genetics*, **2020**, 107, 963-976	11	4
466	Insufficient Evidence for "Autism-Specific" Genes. *American Journal of Human Genetics*, **2020**, 106, 587-595	11	52
465	Implementation of population-based newborn screening reveals low incidence of spinal muscular atrophy. *Genetics in Medicine*, **2020**, 22, 1296-1302	8.1	23
464	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. *Nature Genetics*, **2020**, 52, 572-581	36.3	76
463	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. *Genetics in Medicine*, **2020**, 22, 1215-1226	8.1	7
462	A qualitative study of Latinx parents' experiences of clinical exome sequencing. *Journal of Genetic Counseling*, **2020**, 29, 574-586	2.5	3
461	Choices, attitudes, and experiences of genetic screening in Latino/a and Ashkenazi Jewish individuals. *Journal of Community Genetics*, **2020**, 11, 391-403	2.5	1
460	Returning Results in the Genomic Era: Initial Experiences of the eMERGE Network. *Journal of Personalized Medicine*, **2020**, 10,	3.6	19
459	Frequency of genomic secondary findings among 21,915 eMERGE network participants. *Genetics in Medicine*, **2020**, 22, 1470-1477	8.1	23
458	COVID-19's Impact on Genetics at One Medical Center in New York. *Genetics in Medicine*, **2020**, 22, 1467-1469	8.1	13
457	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	14.6	75
456	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. *Human Mutation*, **2020**, 41, 1577-1587	4.7	4
455	Novel Mutations and Decreased Expression of the Epigenetic Regulator in Pulmonary Arterial Hypertension. *Circulation*, **2020**, 141, 1986-2000	16.7	28

454	Genomic analyses implicate noncoding de novo variants in congenital heart disease. *Nature Genetics*, **2020**, 52, 769-777	36.3	33
453	Evaluating heterogeneity in ASD symptomatology, cognitive ability, and adaptive functioning among 16p11.2 CNV carriers. *Autism Research*, **2020**, 13, 1300-1310	5.1	7
452	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	3.6	14
451	Characterization of the Cancer Spectrum in Men With Germline BRCA1 and BRCA2 Pathogenic Variants: Results From the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *JAMA Oncology*, **2020**, 6, 1218-1230	13.4	25
450	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (KIF1A). *Human Mutation*, **2020**, 41, 1761-1774	4.7	10
449	De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. *Circulation Genomic and Precision Medicine*, **2020**, 13, e002836	5.2	15
448	De novo heterozygous missense and loss-of-function variants in CDC42BPB are associated with a neurodevelopmental phenotype. *American Journal of Medical Genetics, Part A*, **2020**, 182, 962-973	2.5	1
447	Psychiatric and Medical Profiles of Autistic Adults in the SPARK Cohort. *Journal of Autism and Developmental Disorders*, **2020**, 50, 3679-3698	4.6	15
446	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
445	Genetics and Other Omics in Pediatric Pulmonary Arterial Hypertension. *Chest*, **2020**, 157, 1287-1295	5.3	11
444	Phenotypic expansion of Bosch-Boonstra-Schaaf optic atrophy syndrome and further evidence for genotype-phenotype correlations. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1426-1437	2.5	8
443	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. *American Journal of Human Genetics*, **2020**, 106, 467-483	11	12
442	Rapid exome sequencing in PICU patients with new-onset metabolic or neurological disorders. *Pediatric Research*, **2020**, 88, 761-768	3.2	8
441	Differences in brain structure and function in children with the obesity-risk allele. *Obesity Science and Practice*, **2020**, 6, 409-424	2.6	5
440	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
439	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. *PLoS Genetics*, **2020**, 16, e1009189	6	7
438	A Novel On-Site Volunteer Community Infection Prevention Team Prevented Outbreaks at a Hurricane Harvey Mega-Shelter. *Infection Control and Hospital Epidemiology*, **2020**, 41, s100-s100	2	
437	mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. *ELife*, **2020**, 9,	8.9	9

436	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
435	Comparative outcomes of right versus left congenital diaphragmatic hernia: A multicenter analysis. *Journal of Pediatric Surgery*, **2020**, 55, 33-38	2.6	8
434	Psychotic symptoms in 16p11.2 copy-number variant carriers. *Autism Research*, **2020**, 13, 187-198	5.1	4
433	Impact of patient education videos on genetic counseling outcomes after exome sequencing. *Patient Education and Counseling*, **2020**, 103, 127-135	3.1	8
432	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
431	Genetic Basis of Human Congenital Heart Disease. *Cold Spring Harbor Perspectives in Biology*, **2020**, 12,	10.2	11
430	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	3.4	1
429	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. *Nature*, **2020**, 586, 749-756	5.4	122
428	Monogenic variants in dystonia: an exome-wide sequencing study. *Lancet Neurology, The*, **2020**, 19, 908-918	24.1	51
427	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	8.1	34
426	Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1333-1348	5.4	10
425	Language characterization in 16p11.2 deletion and duplication syndromes. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2020**, 183, 380-391	3.5	8
424	Common germline-somatic variant interactions in advanced urothelial cancer. *Nature Communications*, **2020**, 11, 6195	17.4	6
423	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
422	Human iPSC-Derived Neuronal Cells From -Mutated Patients Reveal Altered Expression of Neurodevelopmental Gene Networks. *Frontiers in Neuroscience*, **2020**, 14, 562292	5.1	1
421	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 432-444	11	31
420	Participant choices for return of genomic results in the eMERGE Network. *Genetics in Medicine*, **2020**, 22, 1821-1829	8.1	14
419	Likely damaging de novo variants in congenital diaphragmatic hernia patients are associated with worse clinical outcomes. *Genetics in Medicine*, **2020**, 22, 2020-2028	8.1	8

418	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	4	0
417	Early-Onset Parkinsonism Is a Manifestation of the PPP2R5D p.E200K Mutation. *Annals of Neurology*, **2020**, 88, 1028-1033	9.4	10
416	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
415	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, 100008-100008	0.8	3
414	Genetics and Genomics of Pediatric Pulmonary Arterial Hypertension. *Genes*, **2020**, 11,	4.2	8
413	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	18.1	76
412	Bayesian Inference Associates Rare Variants with Specific Phenotypes in Pulmonary Arterial Hypertension. *Circulation Genomic and Precision Medicine*, **2020**,	5.2	9
411	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	10.1	15
410	Is there a duty to reinterpret genetic data? The ethical dimensions. *Genetics in Medicine*, **2020**, 22, 633-639	8.1	21
409	The influence of genetics in congenital diaphragmatic hernia. *Seminars in Perinatology*, **2020**, 44, 151169	3.3	19
408	A framework for an evidence-based gene list relevant to autism spectrum disorder. *Nature Reviews Genetics*, **2020**, 21, 367-376	30.1	30
407	Genetic attribution and perceived impact of epilepsy in multiplex epilepsy families. *Epilepsia*, **2019**, 60, 2286-2293	6.4	2
406	Familial X-Linked Acrogigantism: Postnatal Outcomes and Tumor Pathology in a Prenatally Diagnosed Infant and His Mother. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 4667-4675	5.6	11
405	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *American Journal of Human Genetics*, **2019**, 105, 588-605	11	63
404	Psychiatric disorders in children with 16p11.2 deletion and duplication. *Translational Psychiatry*, **2019**, 9, 8	8.6	46
403	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2019**, 121, 180-192	8.7	13
402	Identification of a secondary mutation in a pediatric patient with relapsed acute myeloid leukemia leads to the diagnosis and treatment of asymptomatic metastatic medullary thyroid cancer in a parent: a case for sequencing the germline. *Journal of Physical Education and Sports Management*, **2019**, 5,	2.8	2
401	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	11.5	17

400	Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. *Genetics in Medicine*, **2019**, 21, 2413-2421	8.1	164
399	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	8.1	32
398	United States Pulmonary Hypertension Scientific Registry (USPHSR): rationale, design, and clinical implications. *Pulmonary Circulation*, **2019**, 9, 2045894019851696	2.7	5
397	A GENETIC FIRST APPROACH TO DISSECTING THE HETEROGENEITY OF AUTISM: PHENOTYPIC COMPARISON OF AUTISM RISK COPY NUMBER VARIANTS. *European Neuropsychopharmacology*, **2019**, 29, S783-S784	1.2	3
396	Points to consider in the reevaluation and reanalysis of genomic test results: a statement of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*, **2019**, 21, 1267-1270	8.1	59
395	Homozygous noncanonical splice variant in in two siblings with multiple congenital anomalies and global developmental delay. *Journal of Physical Education and Sports Management*, **2019**, 5,	2.8	3
394	Regular use of aspirin and other non-steroidal anti-inflammatory drugs and breast cancer risk for women at familial or genetic risk: a cohort study. *Breast Cancer Research*, **2019**, 21, 52	8.3	29
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5	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1

4	Rare variant analysis of 4,241 pulmonary arterial hypertension cases from an international consortium implicate FBLN2, PDGFD and rare de novo variants in PAH	4
3	Genotype and defects in microtubule-based motility correlate with clinical severity in KIF1A Associated Neurological Disorder	1
2	Whole exome sequencing and characterization of coding variation in 49,960 individuals in the UK Biobank	56
1	Imputing cognitive impairment in SPARK, a large autism cohort	1