Wendy Chung

List of Publications by Citations

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	Strong association of de novo copy number mutations with autism. *Science*, **2007**, 316, 445-9	33.3	2126
560	Induced pluripotent stem cells generated from patients with ALS can be differentiated into motor neurons. *Science*, **2008**, 321, 1218-21	33.3	1572
559	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. *JAMA - Journal of the American Medical Association*, **2017**, 317, 2402-2416	27.4	1140
558	Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. *Genetics in Medicine*, **2017**, 19, 249-255	8.1	1017
557	A copy number variation morbidity map of developmental delay. *Nature Genetics*, **2011**, 43, 838-46	36.3	931
556	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. *Science*, **2015**, 347, 1436-41	33.3	642
555	Pediatric Pulmonary Hypertension: Guidelines From the American Heart Association and American Thoracic Society. *Circulation*, **2015**, 132, 2037-99	16.7	624
554	De novo mutations in histone-modifying genes in congenital heart disease. *Nature*, **2013**, 498, 220-3	50.4	591
553	Clinical application of whole-exome sequencing across clinical indications. *Genetics in Medicine*, **2016**, 18, 696-704	8.1	532
552	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. *Science*, **2015**, 350, 1262-6	33.3	406
551	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. *Nature Genetics*, **2017**, 49, 1593-1601	36.3	348
550	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscular Disorders*, **2018**, 28, 103-115	2.9	319
549	A novel channelopathy in pulmonary arterial hypertension. *New England Journal of Medicine*, **2013**, 369, 351-361	59.2	311
548	Genetics and genomics of pulmonary arterial hypertension. *Journal of the American College of Cardiology*, **2009**, 54, S32-S42	15.1	292
547	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1347-61	27.4	286
546	Return of genomic results to research participants: the floor, the ceiling, and the choices in between. *American Journal of Human Genetics*, **2014**, 94, 818-26	11	283
545	Whole exome sequencing to identify a novel gene (caveolin-1) associated with human pulmonary arterial hypertension. *Circulation: Cardiovascular Genetics*, **2012**, 5, 336-43		268

544	Analysis of GNAS mutations in 60 growth hormone secreting pituitary tumors: correlation with clinical and pathological characteristics and surgical outcome based on highly sensitive GH and IGF-I criteria for remission. *Pituitary*, **2007**, 10, 275-82	4.3	258
543	Observational study of spinal muscular atrophy type I and implications for clinical trials. *Neurology*, **2014**, 83, 810-7	6.5	248
542	Glucocerebrosidase activity in Parkinson's disease with and without GBA mutations. *Brain*, **2015**, 138, 2648-58	11.2	234
541	Genetics and genomics of pulmonary arterial hypertension. *Journal of the American College of Cardiology*, **2013**, 62, D13-21	15.1	228
540	The molecular genetics of rodent single gene obesities. *Journal of Biological Chemistry*, **1997**, 272, 31937-40	5.4	216
539	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
538	Enalapril in infants with single ventricle: results of a multicenter randomized trial. *Circulation*, **2010**, 122, 333-40	16.7	213
537	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209
536	Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics. *Genetics in Medicine*, **2014**, 16, e1	8.1	207
535	BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. *Lancet Respiratory Medicine,the*, **2016**, 4, 129-37	35.1	202
534	MC4R-dependent suppression of appetite by bone-derived lipocalin 2. *Nature*, **2017**, 543, 385-390	50.4	191
533	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
532	Glycogen storage disease type III diagnosis and management guidelines. *Genetics in Medicine*, **2010**, 12, 446-63	8.1	186
531	Genetic Basis for Congenital Heart Disease: Revisited: A Scientific Statement From the American Heart Association. *Circulation*, **2018**, 138, e653-e711	16.7	184
530	A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. *Journal of Medical Genetics*, **2012**, 49, 660-8	5.8	182
529	Genetics and genomics of pulmonary arterial hypertension. *European Respiratory Journal*, **2019**, 53,	13.6	179
528	Pathogenic and likely pathogenic variant prevalence among the first 10,000 patients referred for next-generation cancer panel testing. *Genetics in Medicine*, **2016**, 18, 823-32	8.1	172
527	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. *Neuron*, **2015**, 88, 499-513	13.9	170

526	Pancreatic cancer screening in a prospective cohort of high-risk patients: a comprehensive strategy of imaging and genetics. *Clinical Cancer Research*, **2010**, 16, 5028-37	12.9	167
525	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
524	Copy-number disorders are a common cause of congenital kidney malformations. *American Journal of Human Genetics*, **2012**, 91, 987-97	11	161
523	RASA1 mutations and associated phenotypes in 68 families with capillary malformation-arteriovenous malformation. *Human Mutation*, **2013**, 34, 1632-41	4.7	160
522	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	15.7	158
521	Induced pluripotent stem cells used to reveal drug actions in a long QT syndrome family with complex genetics. *Journal of General Physiology*, **2013**, 141, 61-72	3.4	158
520	A public resource facilitating clinical use of genomes. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 11920-7	11.5	154
519	The usefulness of whole-exome sequencing in routine clinical practice. *Genetics in Medicine*, **2014**, 16, 922-31	8.1	150
518	EIF2AK4 mutations in pulmonary capillary hemangiomatosis. *Chest*, **2014**, 145, 231-236	5.3	143
517	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. *Human Mutation*, **2015**, 36, 1113-27	4.7	142
516	The cognitive and behavioral phenotype of the 16p11.2 deletion in a clinically ascertained population. *Biological Psychiatry*, **2015**, 77, 785-93	7.9	140
515	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
514	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, 343-52	11	136
513	Outcomes of congenital diaphragmatic hernia in the modern era of management. *Journal of Pediatrics*, **2013**, 163, 114-9.e1	3.6	133
512	Comparison of Parkinson risk in Ashkenazi Jewish patients with Gaucher disease and GBA heterozygotes. *JAMA Neurology*, **2014**, 71, 752-7	17.2	132
511	Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling. *Circulation*, **2017**, 136, 1037-1048	16.7	130
510	Progress in Understanding and Treating SCN2A-Mediated Disorders. *Trends in Neurosciences*, **2018**, 41, 442-456	13.3	128
509	Pediatric Cardiomyopathies. *Circulation Research*, **2017**, 121, 855-873	15.7	124

508	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. *Nature*, **2020**, 586, 749-756	556	122
507	Attitudes and practices among internists concerning genetic testing. *Journal of Genetic Counseling*, **2013**, 22, 90-100	2.5	121
506	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
505	Hypomorphism for RPGRIP1L, a ciliary gene vicinal to the FTO locus, causes increased adiposity in mice. *Cell Metabolism*, **2014**, 19, 767-79	24.6	120
504	Genetic testing for dilated cardiomyopathy in clinical practice. *Journal of Cardiac Failure*, **2012**, 18, 296-303	3.3	118
503	Fine structure of the murine leptin receptor gene: splice site suppression is required to form two alternatively spliced transcripts. *Genomics*, **1997**, 45, 264-70	4.3	117
502	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. *American Journal of Human Genetics*, **2015**, 97, 457-64	11	115
501	Observational study of spinal muscular atrophy type 2 and 3: functional outcomes over 1 year. *Archives of Neurology*, **2011**, 68, 779-86		114
500	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. *Neuron*, **2018**, 97, 488-493	13.9	112
499	□cell dysfunction due to increased ER stress in a stem cell model of Wolfram syndrome. *Diabetes*, **2014**, 63, 923-33	0.9	108
498	Opposing brain differences in 16p11.2 deletion and duplication carriers. *Journal of Neuroscience*, **2014**, 34, 11199-211	6.6	108
497	A recurrent PDGFRB mutation causes familial infantile myofibromatosis. *American Journal of Human Genetics*, **2013**, 92, 996-1000	11	108
496	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. *American Journal of Human Genetics*, **2016**, 98, 1051-1066	11	107
495	Cut-like homeobox 1 (CUX1) regulates expression of the fat mass and obesity-associated and retinitis pigmentosa GTPase regulator-interacting protein-1-like (RPGRIP1L) genes and coordinates leptin receptor signaling. *Journal of Biological Chemistry*, **2011**, 286, 2155-70	5.4	105
494	The Congenital Heart Disease Genetic Network Study: rationale, design, and early results. *Circulation Research*, **2013**, 112, 698-706	15.7	104
493	Validation of the Expanded Hammersmith Functional Motor Scale in spinal muscular atrophy type II and III. *Journal of Child Neurology*, **2011**, 26, 1499-507	2.5	102
492	The contribution of de novo and rare inherited copy number changes to congenital heart disease in an unselected sample of children with conotruncal defects or hypoplastic left heart disease. *Human Genetics*, **2014**, 133, 11-27	6.3	96
491	Implementation of next generation sequencing into pediatric hematology-oncology practice: moving beyond actionable alterations. *Genome Medicine*, **2016**, 8, 133	14.4	95

490	Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. *Journal of Clinical Oncology*, **2016**, 34, 2172-81	2.2	95
489	ResearchersPviews on return of incidental genomic research results: qualitative and quantitative findings. *Genetics in Medicine*, **2013**, 15, 888-95	8.1	94
488	Complex genetics and the etiology of human congenital heart disease. *Cold Spring Harbor Perspectives in Medicine*, **2014**, 4, a013953	5.4	93
487	Genetic loss of SH2B3 in acute lymphoblastic leukemia. *Blood*, **2013**, 122, 2425-32	2.2	92
486	Congenital diaphragmatic hernias: from genes to mechanisms to therapies. *DMM Disease Models and Mechanisms*, **2017**, 10, 955-970	4.1	90
485	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	2.5	86
484	Clinical phenotype of the recurrent 1q21.1 copy-number variant. *Genetics in Medicine*, **2016**, 18, 341-9	8.1	84
483	The role of parental cognitive, behavioral, and motor profiles in clinical variability in individuals with chromosome 16p11.2 deletions. *JAMA Psychiatry*, **2015**, 72, 119-26	14.5	84
482	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. *Npj Genomic Medicine*, **2019**, 4, 19	6.2	84
481	Genome-wide association analysis identifies a susceptibility locus for pulmonary arterial hypertension. *Nature Genetics*, **2013**, 45, 518-21	36.3	82
480	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
479	Blood leukocyte microarrays to diagnose systemic onset juvenile idiopathic arthritis and follow the response to IL-1 blockade. *Journal of Experimental Medicine*, **2009**, 206, 2299-2299	16.6	78
478	MSH6 and PMS2 germ-line pathogenic variants implicated in Lynch syndrome are associated with breast cancer. *Genetics in Medicine*, **2018**, 20, 1167-1174	8.1	77
477	iPSC-derived □ells model diabetes due to glucokinase deficiency. *Journal of Clinical Investigation*, **2013**, 123, 3146-53	15.9	77
476	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
475	Congenital disorder of glycosylation id presenting with hyperinsulinemic hypoglycemia and islet cell hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 4371-5	5.6	76
474	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
473	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

472	CANOES: detecting rare copy number variants from whole exome sequencing data. *Nucleic Acids Research*, **2014**, 42, e97	20.1	75
471	10-year performance of four models of breast cancer risk: a validation study. *Lancet Oncology, The*, **2019**, 20, 504-517	21.7	73
470	Effect of copy number variants on outcomes for infants with single ventricle heart defects. *Circulation: Cardiovascular Genetics*, **2013**, 6, 444-51		70
469	Impact of Panel Gene Testing for Hereditary Breast and Ovarian Cancer on Patients. *Journal of Genetic Counseling*, **2017**, 26, 1116-1129	2.5	69
468	Validation of the Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND). *Pediatric Physical Therapy*, **2011**, 23, 322-6	0.9	69
467	Serum endostatin is a genetically determined predictor of survival in pulmonary arterial hypertension. *American Journal of Respiratory and Critical Care Medicine*, **2015**, 191, 208-18	10.2	68
466	Variants in GATA4 are a rare cause of familial and sporadic congenital diaphragmatic hernia. *Human Genetics*, **2013**, 132, 285-92	6.3	67
465	Risk of pancreatic cancer in breast cancer families from the breast cancer family registry. *Cancer Epidemiology Biomarkers and Prevention*, **2013**, 22, 803-11	4	67
464	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. *Genome Medicine*, **2018**, 10, 56	14.4	66
463	Amino acid variants in the human leptin receptor: lack of association to juvenile onset obesity. *Biochemical and Biophysical Research Communications*, **1997**, 233, 248-52	3.4	66
462	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. *Circulation Genomic and Precision Medicine*, **2018**, 11, e001887	5.2	65
461	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
460	Use of genetic tests among neurologists and psychiatrists: knowledge, attitudes, behaviors, and needs for training. *Journal of Genetic Counseling*, **2014**, 23, 156-63	2.5	64
459	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *American Journal of Human Genetics*, **2019**, 105, 588-605	11	63
458	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, 1231-42	8.6	63
457	Glut1 deficiency syndrome and erythrocyte glucose uptake assay. *Annals of Neurology*, **2011**, 70, 996-1005	9.4	63
456	Glut1 deficiency: inheritance pattern determined by haploinsufficiency. *Annals of Neurology*, **2010**, 68, 955-8	9.4	63
455	The genetic basis of pulmonary arterial hypertension. *Human Genetics*, **2014**, 133, 471-9	6.3	62

454	Copy number variants and infantile spasms: evidence for abnormalities in ventral forebrain development and pathways of synaptic function. *European Journal of Human Genetics*, **2011**, 19, 1238-45	5.3	62
453	Absence epilepsy in apathetic, a spontaneous mutant mouse lacking the h channel subunit, HCN2. *Neurobiology of Disease*, **2009**, 33, 499-508	7.5	61
452	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, 485-92	8.1	60
451	Pilot study of population-based newborn screening for spinal muscular atrophy in New York state. *Genetics in Medicine*, **2018**, 20, 608-613	8.1	60
450	Return of secondary genomic findings vs patient autonomy: implications for medical care. *JAMA - Journal of the American Medical Association*, **2013**, 310, 369-70	27.4	60
449	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
448	High prevalence of BRCA1 and BRCA2 germline mutations with loss of heterozygosity in a series of resected pancreatic adenocarcinoma and other neoplastic lesions. *Clinical Cancer Research*, **2013**, 19, 3396-403	12.9	58
447	PVDOMICS: A Multi-Center Study to Improve Understanding of Pulmonary Vascular Disease Through Phenomics. *Circulation Research*, **2017**, 121, 1136-1139	15.7	58
446	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. *American Journal of Human Genetics*, **2019**, 104, 213-228	11	58
445	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. *European Journal of Human Genetics*, **2016**, 24, 652-9	5.3	57
444	Rare variant phasing and haplotypic expression from RNA sequencing with phASER. *Nature Communications*, **2016**, 7, 12817	17.4	57
443	Spectrum of neuropathophysiology in spinal muscular atrophy type I. *Journal of Neuropathology and Experimental Neurology*, **2015**, 74, 15-24	3.1	56
442	De novo copy number variants are associated with congenital diaphragmatic hernia. *Journal of Medical Genetics*, **2012**, 49, 650-9	5.8	56
441	Short communication: the cardiac myosin binding protein C Arg502Trp mutation: a common cause of hypertrophic cardiomyopathy. *Circulation Research*, **2010**, 106, 1549-52	15.7	56
440	Whole exome sequencing and characterization of coding variation in 49,960 individuals in the UK Biobank		56
439	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
438	A complete deficiency of Hyaluronoglucosaminidase 1 (HYAL1) presenting as familial juvenile idiopathic arthritis. *Journal of Inherited Metabolic Disease*, **2011**, 34, 1013-22	5.4	55
437	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. *Biological Psychiatry*, **2019**, 85, 287-297	7.9	55

436	Deep Phenotyping on Electronic Health Records Facilitates Genetic Diagnosis by Clinical Exomes. *American Journal of Human Genetics*, **2018**, 103, 58-73	11	55
435	Genetic evaluation and counseling for epilepsy. *Nature Reviews Neurology*, **2010**, 6, 445-53	15	54
434	Genetic modifiers of Leprfa associated with variability in insulin production and susceptibility to NIDDM. *Genomics*, **1997**, 41, 332-44	4.3	54
433	Decision-making about reproductive choices among individuals at-risk for Huntington's disease. *Journal of Genetic Counseling*, **2007**, 16, 347-62	2.5	54
432	Fatal infantile neuromuscular presentation of glycogen storage disease type IV. *Neuromuscular Disorders*, **2004**, 14, 253-60	2.9	54
431	Aberrant white matter microstructure in children with 16p11.2 deletions. *Journal of Neuroscience*, **2014**, 34, 6214-23	6.6	53
430	Informed consent for return of incidental findings in genomic research. *Genetics in Medicine*, **2014**, 16, 367-73	8.1	53
429	Positional cloning of "Lisch-Like", a candidate modifier of susceptibility to type 2 diabetes in mice. *PLoS Genetics*, **2008**, 4, e1000137	6	53
428	Molecular and functional analysis of SLC25A20 mutations causing carnitine-acylcarnitine translocase deficiency. *Human Mutation*, **2004**, 24, 312-20	4.7	53
427	Insufficient Evidence for "Autism-Specific" Genes. *American Journal of Human Genetics*, **2020**, 106, 587-595	11	52
426	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. *Genetics in Medicine*, **2017**, 19, 575-582	8.1	52
425	Monogenic variants in dystonia: an exome-wide sequencing study. *Lancet Neurology, The*, **2020**, 19, 908-918	24.1	51
424	Advances in the Understanding of the Genetic Determinants of Congenital Heart Disease and Their Impact on Clinical Outcomes. *Journal of the American Heart Association*, **2018**, 7,	6	50
423	Developmental outcomes of children with congenital diaphragmatic hernia: a multicenter prospective study. *Journal of Pediatric Surgery*, **2013**, 48, 1995-2004	2.6	50
422	Defining a comprehensive verotype using electronic health records for personalized medicine. *Journal of the American Medical Informatics Association: JAMIA*, **2013**, 20, e232-8	8.6	50
421	BRCA1 and BRCA2 mutation carriers in the Breast Cancer Family Registry: an open resource for collaborative research. *Breast Cancer Research and Treatment*, **2009**, 116, 379-86	4.4	49
420	Genetic causes of congenital diaphragmatic hernia. *Seminars in Fetal and Neonatal Medicine*, **2014**, 19, 324-30	3.7	48
419	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47

418	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. *PLoS Genetics*, **2018**, 14, e1007822	6	47
417	Psychiatric disorders in children with 16p11.2 deletion and duplication. *Translational Psychiatry*, **2019**, 9, 8	8.6	46
416	Association of plastin 3 expression with disease severity in spinal muscular atrophy only in postpubertal females. *Archives of Neurology*, **2010**, 67, 1252-6		46
415	A novel and lethal de novo LQT-3 mutation in a newborn with distinct molecular pharmacology and therapeutic response. *PLoS ONE*, **2007**, 2, e1258	3.7	46
414	APRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. *Nature Communications*, **2018**, 9, 67	17.4	45
413	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	11	45
412	Combined OXPHOS complex I and IV defect, due to mutated complex I assembly factor C20ORF7. *Journal of Inherited Metabolic Disease*, **2012**, 35, 125-31	5.4	45
411	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. *Genome Medicine*, **2019**, 11, 69	14.4	45
410	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	8.1	45
409	Whole exome sequencing identifies de novo mutations in GATA6 associated with congenital diaphragmatic hernia. *Journal of Medical Genetics*, **2014**, 51, 197-202	5.8	44
408	Increased burden of de novo predicted deleterious variants in complex congenital diaphragmatic hernia. *Human Molecular Genetics*, **2015**, 24, 4764-73	5.6	44
407	Renin-angiotensin-aldosterone genotype influences ventricular remodeling in infants with single ventricle. *Circulation*, **2011**, 123, 2353-62	16.7	44
406	Common variants at the 19p13.1 and ZNF365 loci are associated with ER subtypes of breast cancer and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 645-57	4	44
405	Mutations in SLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. *Journal of Medical Genetics*, **2015**, 52, 541-7	5.8	43
404	The Congenital Heart Disease Genetic Network Study: Cohort description. *PLoS ONE*, **2018**, 13, e0191319	3.7	43
403	Brief Report: SETD2 Mutation in a Child with Autism, Intellectual Disabilities and Epilepsy. *Journal of Autism and Developmental Disorders*, **2015**, 45, 3764-70	4.6	42
402	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotype-phenotype correlations. *Genetics in Medicine*, **2016**, 18, 1143-1150	8.1	42
401	Mutation in SNAP25 as a novel genetic cause of epilepsy and intellectual disability. *Rare Diseases (Austin, Tex)*, **2013**, 1, e26314		42

400	Suggestive linkages between markers on human 1p32-p22 and body fat and insulin levels in the Quebec Family Study. *Obesity*, **1997**, 5, 115-21		42
399	Analysis of significance patterns identifies ubiquitous and disease-specific gene-expression signatures in patient peripheral blood leukocytes. *Annals of the New York Academy of Sciences*, **2005**, 1062, 146-54	6.5	42
398	SMPD1 mutations, activity, and -synuclein accumulation in Parkinson disease. *Movement Disorders*, **2019**, 34, 526-535	7	41
397	Molecular genetic analysis of a human neuropeptide Y receptor. The human homolog of the murine "Y5" receptor may be a pseudogene. *Journal of Biological Chemistry*, **1997**, 272, 3622-7	5.4	41
396	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
395	Frequency of variants in autopsy-proven multiple system atrophy. *Movement Disorders Clinical Practice*, **2017**, 4, 574-581	2.2	40
394	Mutations in ARID2 are associated with intellectual disabilities. *Neurogenetics*, **2015**, 16, 307-14	3	40
393	CDKL5 and ARX mutations in males with early-onset epilepsy. *Pediatric Neurology*, **2013**, 48, 367-77	2.9	40
392	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. *American Journal of Human Genetics*, **2017**, 100, 117-127	11	39
391	BRCA1 and BRCA2 germline mutations are frequently demonstrated in both high-risk pancreatic cancer screening and pancreatic cancer cohorts. *Cancer*, **2014**, 120, 1960-7	6.4	39
390	Genome-wide enrichment of damaging de novo variants in patients with isolated and complex congenital diaphragmatic hernia. *Human Genetics*, **2017**, 136, 679-691	6.3	38
389	Solid phase capturable dideoxynucleotides for multiplex genotyping using mass spectrometry. *Nucleic Acids Research*, **2002**, 30, e85	20.1	38
388	FTO genotype impacts food intake and corticolimbic activation. *American Journal of Clinical Nutrition*, **2018**, 107, 145-154	7	37
387	Natural history and genotype-phenotype correlations in 72 individuals with SATB2-associated syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 925-935	2.5	37
386	Cohort Profile: The Breast Cancer Prospective Family Study Cohort (ProF-SC). *International Journal of Epidemiology*, **2016**, 45, 683-92	7.8	37
385	Mutations in COQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. *Journal of Medical Genetics*, **2015**, 52, 627-35	5.8	36
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