Stephen Scherer

List of Publications by Year
in descending order

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663	97,920	333 137	339 285
papers	citations	h-index	g-index

725	725	725	84323
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Global variation in copy number in the human genome. Nature, 2006, 444, 444-454.	13.7	3,831
2	An atlas of genetic correlations across human diseases and traits. Nature Genetics, 2015, 47, 1236-1241.	9.4	3,145
3	Detection of large-scale variation in the human genome. Nature Genetics, 2004, 36, 949-951.	9.4	2,602
4	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. American Journal of Human Genetics, 2010, 86, 749-764.	2.6	2,325
5	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
6	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
7	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
8	Structural variation in the human genome. Nature Reviews Genetics, 2006, 7, 85-97.	7.7	1,745
9	Origins and functional impact of copy number variation in the human genome. Nature, 2010, 464, 704-712.	13.7	1,721
10	Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488.	2.6	1,641
11	Mutations in ABC1 in Tangier disease and familial high-density lipoprotein deficiency. Nature Genetics, 1999, 22, 336-345.	9.4	1,609
12	Relative Impact of Nucleotide and Copy Number Variation on Gene Expression Phenotypes. Science, 2007, 315, 848-853.	6.0	1,546
13	The Diploid Genome Sequence of an Individual Human. PLoS Biology, 2007, 5, e254.	2.6	1,491
14	Germline and somatic mutations in the tyrosine kinase domain of the MET proto-oncogene in papillary renal carcinomas. Nature Genetics, 1997, 16, 68-73.	9.4	1,461
15	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
16	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	9.4	1,272
17	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
18	Mutations in the human Sonic Hedgehog gene cause holoprosencephaly. Nature Genetics, 1996, 14, 357-360.	9.4	1,075

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19	The human splicing code reveals new insights into the genetic determinants of disease. Science, 2015, 347, 1254806.	6.0	1,053
20	The Database of Genomic Variants: a curated collection of structural variation in the human genome. Nucleic Acids Research, 2014, 42, D986-D992.	6.5	1,033
21	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	9.4	838
22	MADR2 Maps to 18q21 and Encodes a TGFÎ²â€“Regulated MADâ€“Related Protein That Is Functionally Mutated in Colorectal Carcinoma. Cell, 1996, 86, 543-552.	13.5	833
23	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
24	Subgroup-specific structural variation across 1,000 medulloblastoma genomes. Nature, 2012, 488, 49-56.	13.7	761
25	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	13.7	737
26	Mutations in SUFU predispose to medulloblastoma. Nature Genetics, 2002, 31, 306-310.	9.4	722
27	A copy number variation map of the human genome. Nature Reviews Genetics, 2015, 16, 172-183.	7.7	707
28	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	7.1	701
29	Copy number variation: New insights in genome diversity. Genome Research, 2006, 16, 949-961.	2.4	697
30	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	7.1	691
31	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	9.4	641
32	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. Nature Genetics, 2001, 29, 166-173.	9.4	635
33	Identification of Sonic hedgehog as a candidate gene responsible for holoprosencephaly. Nature Genetics, 1996, 14, 353-356.	9.4	621
34	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	9.4	621
35	Contribution of SHANK3 Mutations to Autism Spectrum Disorder. American Journal of Human Genetics, 2007, 81, 1289-1297.	2.6	604
36	Structure and chromosomal localization of the human constitutive endothelial nitric oxide synthase gene. Journal of Biological Chemistry, 1993, 268, 17478-88.	1.6	590

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37	Copy-number variations associated with neuropsychiatric conditions. Nature, 2008, 455, 919-923.	13.7	587
38	A genome-wide linkage and association scan reveals novel loci for autism. Nature, 2009, 461, 802-808.	13.7	570
39	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	1.4	538
40	Epigenomic alterations define lethal CIMP-positive ependymomas of infancy. Nature, 2014, 506, 445-450.	13.7	521
41	Cone-Rod Dystrophy Due to Mutations in a Novel Photoreceptor-Specific Homeobox Gene (CRX) Essential for Maintenance of the Photoreceptor. Cell, 1997, 91, 543-553.	13.5	520
42	Identifying Signatures of Natural Selection in Tibetan and Andean Populations Using Dense Genome Scan Data. PLoS Genetics, 2010, 6, e1001116.	1.5	508
43	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	1.5	501
44	Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. Nature Genetics, 1998, 20, 171-174.	9.4	499
45	Meta-analysis of GWAS of over 16,000 individuals with autism spectrum disorder highlights a novel locus at 10q24.32 and a significant overlap with schizophrenia. Molecular Autism, 2017, 8, 21.	2.6	495
46	Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation. Nature Genetics, 2010, 42, 489-491.	9.4	491
47	Whole-genome sequencing of quartet families with autism spectrum disorder. Nature Medicine, 2015, 21, 185-191.	15.2	457
48	Genetic architecture in autism spectrum disorder. Current Opinion in Genetics and Development, 2012, 22, 229-237.	1.5	445
49	Detection of Clinically Relevant Genetic Variants in Autism Spectrum Disorder by Whole-Genome Sequencing. American Journal of Human Genetics, 2013, 93, 249-263.	2.6	429
50	Disruption of a long-range cis-acting regulator for Shh causes preaxial polydactyly. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 7548-7553.	3.3	418
51	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. American Journal of Psychiatry, 2017, 174, 850-858.	4.0	410
52	Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in Medicine, 2018, 20, 435-443.	1.1	404
53	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. Nature Genetics, 2017, 49, 978-985.	9.4	401
54	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394

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55	The gene mutated in adult-onset type II citrullinaemia encodes a putative mitochondrial carrier protein. Nature Genetics, 1999, 22, 159-163.	9.4	392
56	Multiple recurrent genetic events converge on control of histone lysine methylation in medulloblastoma. Nature Genetics, 2009, 41, 465-472.	9.4	391
57	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. Nature Biotechnology, 2011, 29, 512-520.	9.4	384
58	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.	1.1	378
59	Clonal selection drives genetic divergence of metastatic medulloblastoma. Nature, 2012, 482, 529-533.	13.7	376
60	Cloning and mapping of a cDNA for methionine synthase reductase, a flavoprotein defective in patients with homocystinuria. Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 3059-3064.	3.3	371
61	Mutations in ATP6N1B, encoding a new kidney vacuolar proton pump 116-kD subunit, cause recessive distal renal tubular acidosis with preserved hearing. Nature Genetics, 2000, 26, 71-75.	9.4	368
62	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. PLoS Genetics, 2012, 8, e1002521.	1.5	358
63	Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. JAMA - Journal of the American Medical Association, 2015, 314, 895.	3.8	352
64	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	1.4	334
65	Challenges and standards in integrating surveys of structural variation. Nature Genetics, 2007, 39, S7-S15.	9.4	331
66	Identification of C7orf11 (TTDN1) Gene Mutations and Genetic Heterogeneity in Nonphotosensitive Trichothiodystrophy. American Journal of Human Genetics, 2005, 76, 510-516.	2.6	329
67	Single cell-derived clonal analysis of human glioblastoma links functional and genomic heterogeneity. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 851-856.	3.3	321
68	A 1.5 millionâ€“base pair inversion polymorphism in families with Williams-Beuren syndrome. Nature Genetics, 2001, 29, 321-325.	9.4	314
69	Combined hereditary and somatic mutations of replication error repair genes result in rapid onset of ultra-hypermutated cancers. Nature Genetics, 2015, 47, 257-262.	9.4	306
70	Rare Copy Number Variation Discovery and Cross-Disorder Comparisons Identify Risk Genes for ADHD. Science Translational Medicine, 2011, 3, 95ra75.	5.8	304
71	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	1.7	295
72	Mutations in NHLRC1 cause progressive myoclonus epilepsy. Nature Genetics, 2003, 35, 125-127.	9.4	294

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73	SHANK1 Deletions in Males with Autism Spectrum Disorder. American Journal of Human Genetics, 2012, 90, 879-887.	2.6	292
74	GATA1 mutations in transient leukemia and acute megakaryoblastic leukemia of Down syndrome. Blood, 2003, 101, 4301-4304.	0.6	291
75	A previously unidentified MECP2 open reading frame defines a new protein isoform relevant to Rett syndrome. Nature Genetics, 2004, 36, 339-341.	9.4	290
76	A homeobox gene, HLXB9, is the major locus for dominantly inherited sacral agenesis. Nature Genetics, 1998, 20, 358-361.	9.4	287
77	A genome-wide association study of anorexia nervosa. Molecular Psychiatry, 2014, 19, 1085-1094.	4.1	282
78	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	9.4	281
79	Severe Expressive-Language Delay Related to Duplication of the Williamsâ€“Beuren Locus. New England Journal of Medicine, 2005, 353, 1694-1701.	13.9	269
80	Fusion of two novel genes, RBM15 and MKL1, in the t(1;22)(p13;q13) of acute megakaryoblastic leukemia. Nature Genetics, 2001, 28, 220-221.	9.4	268
81	A role for common fragile site induction in amplification of human oncogenes. Cancer Cell, 2002, 1, 89-97.	7.7	267
82	Cytogenetic Prognostication Within Medulloblastoma Subgroups. Journal of Clinical Oncology, 2014, 32, 886-896.	0.8	263
83	Towards a comprehensive structural variation map of an individual human genome. Genome Biology, 2010, 11, R52.	13.9	261
84	Molecular Cloning of the Human Gene, PNKP, Encoding a Polynucleotide Kinase 3â€²-Phosphatase and Evidence for Its Role in Repair of DNA Strand Breaks Caused by Oxidative Damage. Journal of Biological Chemistry, 1999, 274, 24176-24186.	1.6	258
85	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
86	Phenotypic spectrum associated with de novo and inherited deletions and duplications at 16p11.2 in individuals ascertained for diagnosis of autism spectrum disorder. Journal of Medical Genetics, 2010, 47, 195-203.	1.5	244
87	Prepublication data sharing. Nature, 2009, 461, 168-170.	13.7	243
88	Genome-Wide Analysis of Copy Number Variants in Attention Deficit Hyperactivity Disorder: The Role of Rare Variants and Duplications at 15q13.3. American Journal of Psychiatry, 2012, 169, 195-204.	4.0	242
89	Structural variants: changing the landscape of chromosomes and design of disease studies. Human Molecular Genetics, 2006, 15, R57-R66.	1.4	235
90	Hotspots for copy number variation in chimpanzees and humans. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 8006-8011.	3.3	231

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91	Expression Analysis of Six Paralogous Human Hyaluronidase Genes Clustered on Chromosomes 3p21 and 7q31. Genomics, 1999, 60, 356-361.	1.3	229
92	Molecular Analysis of the Pds Gene in Pendred Syndrome (Sensorineural Hearing Loss and Goitre). Human Molecular Genetics, 1998, 7, 1105-1112.	1.4	226
93	Joint Analysis of Psychiatric Disorders Increases Accuracy of Risk Prediction for Schizophrenia, Bipolar Disorder, and Major Depressive Disorder. American Journal of Human Genetics, 2015, 96, 283-294.	2.6	225
94	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	2.6	219
95	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. Nature Neuroscience, 2017, 20, 1217-1224.	7.1	212
96	Molecular Basis for Expression of Common and Rare Fragile Sites. Molecular and Cellular Biology, 2003, 23, 7143-7151.	1.1	211
97	Genome-wide detection of segmental duplications and potential assembly errors in the human genome sequence. Genome Biology, 2003, 4, R25.	13.9	207
98	Frequent Amplification of a chr19q13.41 MicroRNA Polycistron in Aggressive Primitive Neuroectodermal Brain Tumors. Cancer Cell, 2009, 16, 533-546.	7.7	207
99	Copy-number variation in control population cohorts. Human Molecular Genetics, 2007, 16, R168-R173.	1.4	203
100	Molecular characterization of a common fragile site (FRA7H) on human chromosome 7 by the cloning of a simian virus 40 integration site. Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 8141-8146.	3.3	201
101	Genome-wide characteristics of de novo mutations in autism. Npj Genomic Medicine, 2016, 1, 160271-1602710.	1.7	200
102	Excessive genomic DNA copy number variation in the Liâ€“Fraumeni cancer predisposition syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 11264-11269.	3.3	192
103	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	1.1	191
104	Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. American Journal of Human Genetics, 2012, 90, 925-933.	2.6	188
105	Development of bioinformatics resources for display and analysis of copy number and other structural variants in the human genome. Cytogenetic and Genome Research, 2006, 115, 205-214.	0.6	187
106	Human Chromosome 7: DNA Sequence and Biology. Science, 2003, 300, 767-772.	6.0	185
107	Characterization of the split hand/split foot malformation locus SHFM1 at 7q21.3-q22.1 and analysis of a candidate gene for its expression during limb development. Human Molecular Genetics, 1996, 5, 571-579.	1.4	184
108	Rare Deletions at the Neurexin 3 Locus in Autism Spectrum Disorder. American Journal of Human Genetics, 2012, 90, 133-141.	2.6	182

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109	Copy Number Variations in Schizophrenia: Critical Review and New Perspectives on Concepts of Genetics and Disease. American Journal of Psychiatry, 2010, 167, 899-914.	4.0	180
110	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	1.8	180
111	Haploinsufficiency of a Spliceosomal GTPase Encoded by EFTUD2 Causes Mandibulofacial Dysostosis with Microcephaly. American Journal of Human Genetics, 2012, 90, 369-377.	2.6	180
112	Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. Nature Genetics, 2010, 42, 400-405.	9.4	179
113	Mutations in the C-Terminal Domain of Sonic Hedgehog Cause Holoprosencephaly. Human Molecular Genetics, 1997, 6, 1847-1853.	1.4	178
114	Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68.	5.8	178
115	Mutations in NMNAT1 cause Leber congenital amaurosis and identify a new disease pathway for retinal degeneration. Nature Genetics, 2012, 44, 1035-1039.	9.4	177
116	Localization of the gene encoding the Î±2/Î´-subunits of the L-type voltage-dependent calcium channel to chromosome 7q and analysis of the segregation of flanking markers in malignant hyperthermia susceptible families. Human Molecular Genetics, 1994, 3, 969-975.	1.4	176
117	Genome-wide detection of human copy number variations using high-density DNA oligonucleotide arrays. Genome Research, 2006, 16, 1575-1584.	2.4	175
118	A Discovery Resource of Rare Copy Number Variations in Individuals with Autism Spectrum Disorder. G3: Genes, Genomes, Genetics, 2012, 2, 1665-1685.	0.8	175
119	Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331.	6.0	174
120	Disruption of a Novel Gene (IMMP2L) by a Breakpoint in 7q31 Associated with Tourette Syndrome. American Journal of Human Genetics, 2001, 68, 848-858.	2.6	171
121	Absence of a Paternally Inherited FOXP2 Gene in Developmental Verbal Dyspraxia. American Journal of Human Genetics, 2006, 79, 965-972.	2.6	170
122	Cloning and Characterization of on 7q21.3 Encoding a Fourth Pyruvate Dehydrogenase Kinase Isoenzyme in Human. Journal of Biological Chemistry, 1996, 271, 22376-22382.	1.6	167
123	CHARGE and Kabuki Syndromes: Gene-Specific DNA Methylation Signatures Identify Epigenetic Mechanisms Linking These Clinically Overlapping Conditions. American Journal of Human Genetics, 2017, 100, 773-788.	2.6	166
124	Gene and miRNA expression profiles in autism spectrum disorders. Brain Research, 2011, 1380, 85-97.	1.1	165
125	Human PON2 gene at 7q21.3: cloning, multiple mRNA forms, and missense polymorphisms in the coding sequence. Gene, 1998, 213, 149-157.	1.0	163
126	Characterization of the Gene Encoding Human Sarcolipin (SLN), a Proteolipid Associated with SERCA1: Absence of Structural Mutations in Five Patients with Brody Disease. Genomics, 1997, 45, 541-553.	1.3	159

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127	Autism spectrum disorder: advances in evidence-based practice. Cmaj, 2014, 186, 509-519.	0.9	158
128	The clinical context of copy number variation in the human genome. Expert Reviews in Molecular Medicine, 2010, 12, e8.	1.6	157
129	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	2.6	157
130	A novel recurrent mutation in ATP1A3 causes CAPOS syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 15.	1.2	157
131	Expanded Repeat in Canine Epilepsy. Science, 2005, 307, 81-81.	6.0	156
132	Mutations in PIK3R1 Cause SHORT Syndrome. American Journal of Human Genetics, 2013, 93, 158-166.	2.6	156
133	A Comprehensive Workflow for Read Depth-Based Identification of Copy-Number Variation from Whole-Genome Sequence Data. American Journal of Human Genetics, 2018, 102, 142-155.	2.6	156
134	Copy number variations and risk for schizophrenia in 22q11.2 deletion syndrome. Human Molecular Genetics, 2008, 17, 4045-4053.	1.4	155
135	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	13.7	155
136	The Autism Sequencing Consortium: Large-Scale, High-Throughput Sequencing in Autism Spectrum Disorders. Neuron, 2012, 76, 1052-1056.	3.8	153
137	Autism spectrum disorder in the genetics clinic: a review. Clinical Genetics, 2013, 83, 399-407.	1.0	153
138	Genome assembly comparison identifies structural variants in the human genome. Nature Genetics, 2006, 38, 1413-1418.	9.4	150
139	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. PLoS Medicine, 2016, 13, e1001976.	3.9	150
140	Discovery of Human Inversion Polymorphisms by Comparative Analysis of Human and Chimpanzee DNA Sequence Assemblies. PLoS Genetics, 2005, 1, e56.	1.5	149
141	Rare Copy Number Variations in Adults with Tetralogy of Fallot Implicate Novel Risk Gene Pathways. PLoS Genetics, 2012, 8, e1002843.	1.5	149
142	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. Nature Genetics, 2014, 46, 742-747.	9.4	149
143	Ankrd11 Is a Chromatin Regulator Involved in Autism that Is Essential for Neural Development. Developmental Cell, 2015, 32, 31-42.	3.1	147
144	Phase Separation as a Missing Mechanism for Interpretation of Disease Mutations. Cell, 2020, 183, 1742-1756.	13.5	147

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145	Novel glycogen synthase kinase 3 and ubiquitination pathways in progressive myoclonus epilepsy. Human Molecular Genetics, 2005, 14, 2727-2736.	1.4	146
146	Mutation of the CLN6 Gene in Teenage-Onset Progressive Myoclonus Epilepsy. Pediatric Neurology, 2012, 47, 205-208.	1.0	143
147	Germline Mutation of RPS20, Encoding a Ribosomal Protein, Causes Predisposition to Hereditary Nonpolyposis Colorectal Carcinoma Without DNA Mismatch Repair Deficiency. Gastroenterology, 2014, 147, 595-598.e5.	0.6	143
148	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768.	1.4	140
149	Rare exonic deletions implicate the synaptic organizer Gephyrin (GPHN) in risk for autism, schizophrenia and seizures. Human Molecular Genetics, 2013, 22, 2055-2066.	1.4	139
150	HLA-DRB1*11and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 15970-15975.	3.3	139
151	Dysregulation of cyclin dependent kinase 6 expression in splenic marginal zone lymphoma through chromosome 7q translocations. Oncogene, 1999, 18, 6271-6277.	2.6	137
152	Involvement of the HLXB9 Homeobox Gene in Currarino Syndrome. American Journal of Human Genetics, 2000, 66, 312-319.	2.6	137
153	Accurate and reliable high-throughput detection of copy number variation in the human genome. Genome Research, 2006, 16, 1566-1574.	2.4	136
154	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	2.6	135
155	Inherited and de novo SHANK2 variants associated with autism spectrum disorder impair neuronal morphogenesis and physiology. Human Molecular Genetics, 2012, 21, 344-357.	1.4	133
156	Mutation spectrum and predicted function of laforin in Laforaâ€™s progressive myoclonus epilepsy. Neurology, 2000, 55, 341-346.	1.5	131
157	Length of Uninterrupted CAG, Independent of Polyglutamine Size, Results in Increased Somatic Instability, Hastening Onset of Huntington Disease. American Journal of Human Genetics, 2019, 104, 1116-1126.	2.6	130
158	Altered TAOK2 activity causes autism-related neurodevelopmental and cognitive abnormalities through RhoA signaling. Molecular Psychiatry, 2019, 24, 1329-1350.	4.1	128
159	Germ-line DNA copy number variation frequencies in a large North American population. Human Genetics, 2007, 122, 345-353.	1.8	127
160	Molecular Cytogenetic Delineation of Deletions and Translocations Involving Chromosome Band 7q22 in Myeloid Leukemias. Blood, 1997, 89, 2036-2041.	0.6	126
161	Physical mapping of the split hand/split foot locus on chromosome 7 and implication in syndromic ectrodactyly. Human Molecular Genetics, 1994, 3, 1345-1354.	1.4	125
162	Post-transcriptional Regulation of Endothelial Nitric-oxide Synthase by an Overlapping Antisense mRNA Transcript. Journal of Biological Chemistry, 2004, 279, 37982-37996.	1.6	125

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163	Genetic architecture distinguishes systemic juvenile idiopathic arthritis from other forms of juvenile idiopathic arthritis: clinical and therapeutic implications. Annals of the Rheumatic Diseases, 2017, 76, 906-913.	0.5	123
164	Enrichment of segmental duplications in regions of breaks of synteny between the human and mouse genomes suggest their involvement in evolutionary rearrangements. Human Molecular Genetics, 2003, 12, 2201-2208.	1.4	121
165	Rearrangement bursts generate canonical gene fusions in bone and soft tissue tumors. Science, 2018, 361, .	6.0	121
166	Pathogenic rare copy number variants in community-based schizophrenia suggest a potential role for clinical microarrays. Human Molecular Genetics, 2013, 22, 4485-4501.	1.4	120
167	Clinically relevant copy number variations detected in cerebral palsy. Nature Communications, 2015, 6, 7949.	5.8	120
168	Rare Copy Number Variants Contribute to Congenital Left-Sided Heart Disease. PLoS Genetics, 2012, 8, e1002903.	1.5	119
169	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	3.3	118
170	Mutations in C5ORF42 Cause Joubert Syndrome in the French Canadian Population. American Journal of Human Genetics, 2012, 90, 693-700.	2.6	118
171	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	9.4	118
172	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	1.7	118
173	p200 ARF-GEP1: A Golgi-localized guanine nucleotide exchange protein whose Sec7 domain is targeted by the drug brefeldin A. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 7968-7973.	3.3	117
174	Progress in the genetics of autism spectrum disorder. Developmental Medicine and Child Neurology, 2018, 60, 445-451.	1.1	116
175	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. Genome Biology, 2020, 21, 102.	3.8	114
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