Stephen W Scherer

List of Publications by Year
in descending order

308	31,946	9786 73	5255 165
papers	citations	h-index	g-index

318	318	318	41845
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Detection of large-scale variation in the human genome. Nature Genetics, 2004, 36, 949-951.	21.4	2,602
2	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.	6.2	2,325
3	Germline and somatic mutations in the tyrosine kinase domain of the MET proto-oncogene in papillary renal carcinomas. Nature Genetics, 1997, 16, 68-73.	21.4	1,461
4	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
5	Mutations in the human Sonic Hedgehog gene cause holoprosencephaly. Nature Genetics, 1996, 14, 357-360.	21.4	1,075
6	The human splicing code reveals new insights into the genetic determinants of disease. Science, 2015, 347, 1254806.	12.6	1,053
7	The Database of Genomic Variants: a curated collection of structural variation in the human genome. Nucleic Acids Research, 2014, 42, D986-D992.	14.5	1,033
8	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	21.4	838
9	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	6.2	819
10	A copy number variation map of the human genome. Nature Reviews Genetics, 2015, 16, 172-183.	16.3	707
11	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	14.8	691
12	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	21.4	641
13	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. Nature Genetics, 2001, 29, 166-173.	21.4	635
14	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
15	Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. Nature Genetics, 1998, 20, 171-174.	21.4	499
16	Whole-genome sequencing of quartet families with autism spectrum disorder. Nature Medicine, 2015, 21, 185-191.	30.7	457
17	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.	2.4	404
18	The gene mutated in adult-onset type II citrullinaemia encodes a putative mitochondrial carrier protein. Nature Genetics, 1999, 22, 159-163.	21.4	392

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19	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.	2.4	378
20	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.	21.4	368
21	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.	7.4	352
22	Challenges and standards in integrating surveys of structural variation. Nature Genetics, 2007, 39, S7-S15.	21.4	331
23	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.	7.1	321
24	Combined hereditary and somatic mutations of replication error repair genes result in rapid onset of ultra-hypermutated cancers. Nature Genetics, 2015, 47, 257-262.	21.4	306
25	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
26	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	21.4	281
27	Reliability of the CMT neuropathy score (second version) in Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2011, 16, 191-198.	3.1	269
28	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.	21.4	268
29	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	6.2	219
30	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. Nature Neuroscience, 2017, 20, 1217-1224.	14.8	212
31	Genome-wide characteristics of de novo mutations in autism. Npj Genomic Medicine, 2016, 1, 160271-1602710.	3.8	200
32	Human Chromosome 7: DNA Sequence and Biology. Science, 2003, 300, 767-772.	12.6	185
33	A Discovery Resource of Rare Copy Number Variations in Individuals with Autism Spectrum Disorder. G3: Genes, Genomes, Genetics, 2012, 2, 1665-1685.	1.8	175
34	Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331.	12.6	174
35	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.	6.2	166
36	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.	6.2	156

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37	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	27.8	155
38	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. Nature Genetics, 2014, 46, 742-747.	21.4	149
39	Ankrd11 Is a Chromatin Regulator Involved in Autism that Is Essential for Neural Development. Developmental Cell, 2015, 32, 31-42.	7.0	147
40	Phase Separation as a Missing Mechanism for Interpretation of Disease Mutations. Cell, 2020, 183, 1742-1756.	28.9	147
41	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.	1.3	143
42	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.	7.1	139
43	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.	6.2	130
44	Altered TAOK2 activity causes autism-related neurodevelopmental and cognitive abnormalities through RhoA signaling. Molecular Psychiatry, 2019, 24, 1329-1350.	7.9	128
45	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.	0.9	123
46	Rearrangement bursts generate canonical gene fusions in bone and soft tissue tumors. Science, 2018, 361, .	12.6	121
47	Clinically relevant copy number variations detected in cerebral palsy. Nature Communications, 2015, 6, 7949.	12.8	120
48	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	3.8	118
49	PMP22 antisense oligonucleotides reverse Charcot-Marie-Tooth disease type 1A features in rodent models. Journal of Clinical Investigation, 2017, 128, 359-368.	8.2	117
50	Progress in the genetics of autism spectrum disorder. Developmental Medicine and Child Neurology, 2018, 60, 445-451.	2.1	116
51	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. Genome Biology, 2020, 21, 102.	8.8	114
52	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	1.3	114
53	Central nervous system dysfunction in a mouse model of Fa2h deficiency. Glia, 2011, 59, 1009-1021.	4.9	112
54	Copy Number Variation in Obsessive-Compulsive Disorder and Tourette Syndrome: A Cross-Disorder Study. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 910-919.	0.5	111

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55	Complete Disruption of Autism-Susceptibility Genes by Gene Editing Predominantly Reduces Functional Connectivity of Isogenic Human Neurons. Stem Cell Reports, 2018, 11, 1211-1225.	4.8	111
56	Expression of multiple neuregulin transcripts in postnatal rat brains. Journal of Comparative Neurology, 1994, 349, 389-400.	1.6	106
57	Biological Overlap of Attention-Deficit/Hyperactivity Disorder and Autism Spectrum Disorder: Evidence From Copy Number Variants. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 761-770.e26.	0.5	105
58	Syndromic autism spectrum disorders: moving from a clinically defined to a molecularly defined approach. Dialogues in Clinical Neuroscience, 2017, 19, 353-371.	3.7	105
59	Compound heterozygous mutations in the noncoding RNU4ATAC cause Roifman Syndrome by disrupting minor intron splicing. Nature Communications, 2015, 6, 8718.	12.8	104
60	Biallelic mutations in DNAJC21 cause Shwachman-Diamond syndrome. Blood, 2017, 129, 1557-1562.	1.4	104
61	Axons modulate the expression of transforming growth factor-betas in Schwann cells. Glia, 1993, 8, 265-276.	4.9	103
62	Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. Genetics in Medicine, 2015, 17, 149-157.	2.4	103
63	Colorectal carcinomas in mice lacking the catalytic subunit of PI(3)KÎ³. Nature, 2000, 406, 897-902.	27.8	102
64	Risk factors for autism: translating genomic discoveries into diagnostics. Human Genetics, 2011, 130, 123-148.	3.8	102
65	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	21.4	97
66	Physical mapping of the holoprosencephaly critical region on chromosome 7q36. Nature Genetics, 1993, 3, 247-251.	21.4	95
67	Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. Brain, 2015, 138, 845-861.	7.6	94
68	A novel recessive Nefl mutation causes a severe, earlyâ€onset axonal neuropathy. Annals of Neurology, 2009, 66, 759-770.	5.3	90
69	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1912-1918.	30.7	90
70	Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. European Journal of Human Genetics, 2018, 26, 740-744.	2.8	88
71	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
72	De novo and rare inherited copy-number variations in the hemiplegic form of cerebral palsy. Genetics in Medicine, 2018, 20, 172-180.	2.4	82

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73	OTUD7A Regulates Neurodevelopmental Phenotypes in the 15q13.3 Microdeletion Syndrome. American Journal of Human Genetics, 2018, 102, 278-295.	6.2	81
74	Loss of heterozygosity and reduced expression of the CUTL1 gene in uterine leiomyomas. Oncogene, 1997, 14, 2355-2365.	5.9	80
75	Neurofascin antibodies in autoimmune, genetic, and idiopathic neuropathies. Neurology, 2018, 90, e31-e38.	1.1	78
76	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	7.2	77
77	Paraoxonase-2 Gene (PON2) G148 Variant Associated with Elevated Fasting Plasma Glucose in Noninsulin-Dependent Diabetes Mellitus1. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 3373-3377.	3.6	75
78	A high-resolution copy-number variation resource for clinical and population genetics. Genetics in Medicine, 2015, 17, 747-752.	2.4	73
79	CNTN5-/+or EHMT2-/+human iPSC-derived neurons from individuals with autism develop hyperactive neuronal networks. ELife, 2019, 8, .	6.0	72
80	CHD2 haploinsufficiency is associated with developmental delay, intellectual disability, epilepsy and neurobehavioural problems. Journal of Neurodevelopmental Disorders, 2014, 6, 9.	3.1	71
81	Bone marrow failure and developmental delay caused by mutations in poly(A)-specific ribonuclease (PARN). Journal of Medical Genetics, 2015, 52, 738-748.	3.2	71
82	Molecular characterization of NRXN1 deletions from 19,263 clinical microarray cases identifies exons important for neurodevelopmental disease expression. Genetics in Medicine, 2017, 19, 53-61.	2.4	70
83	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
84	15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693.	2.5	67
85	Lethal Disorder of Mitochondrial Fission Caused by Mutations in DNM1L. Journal of Pediatrics, 2016, 171, 313-316.e2.	1.8	67
86	Delineation of multiple deleted regions in 7q in myeloid disorders. Genes Chromosomes and Cancer, 1999, 25, 384-392.	2.8	65
87	Myopathy associated BAG3 mutations lead to protein aggregation by stalling Hsp70 networks. Nature Communications, 2018, 9, 5342.	12.8	65
88	Molecular specializations at nodes and paranodes in peripheral nerve. , 1996, 34, 452-461.		64
89	Intrathecal gene therapy rescues a model of demyelinating peripheral neuropathy. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E2421-9.	7.1	63
90	Structural neuroimaging correlates of social deficits are similar in autism spectrum disorder and attention-deficit/hyperactivity disorder: analysis from the POND Network. Translational Psychiatry, 2019, 9, 72.	4.8	63

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91	A microcosting and costâ€“consequence analysis of clinical genomic testing strategies in autism spectrum disorder. Genetics in Medicine, 2017, 19, 1268-1275.	2.4	62
92	t(7;12)(q36;p13), a new recurrent translocation involvingETV6 in infant leukemia. Genes Chromosomes and Cancer, 2000, 29, 325-332.	2.8	60
93	Nodes, paranodes and neuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 61-71.	1.9	60
94	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	6.2	59
95	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
96	Remote Ischemic Preconditioning in Children Undergoing Cardiac Surgery With Cardiopulmonary Bypass: A Singleâ€Center Doubleâ€Blinded Randomized Trial. Journal of the American Heart Association, 2014, 3, .	3.7	58
97	PMPCAmutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517.	7.6	58
98	Copy Number Variable MicroRNAs in Schizophrenia and Their Neurodevelopmental Gene Targets. Biological Psychiatry, 2015, 77, 158-166.	1.3	58
99	Haploinsufficiency of vascular endothelial growth factor related signaling genes is associated with tetralogy of Fallot. Genetics in Medicine, 2019, 21, 1001-1007.	2.4	58
100	Gene structure of the human MET proto-oncogene. Oncogene, 1997, 15, 1583-1586.	5.9	57
101	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136.	2.0	57
102	Synaptic Dysfunction in Human Neurons With Autism-Associated Deletions in PTCHD1-AS. Biological Psychiatry, 2020, 87, 139-149.	1.3	57
103	Copy number variation in Han Chinese individuals with autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2014, 6, 34.	3.1	55
104	Uncovering obsessive-compulsive disorder risk genes in a pediatric cohort by high-resolution analysis of copy number variation. Journal of Neurodevelopmental Disorders, 2016, 8, 36.	3.1	55
105	Whole-Exome Sequencing and Targeted Copy Number Analysis in Primary Ciliary Dyskinesia. G3: Genes, Genomes, Genetics, 2015, 5, 1775-1781.	1.8	53
106	Axonal pathology in demyelinating diseases. Annals of Neurology, 1999, 45, 6-7.	5.3	52
107	Laforin is a cell membrane and endoplasmic reticulum-associated protein tyrosine phosphatase. Annals of Neurology, 2001, 49, 271-275.	5.3	52
108	Target-enrichment sequencing and copy number evaluation in inherited polyneuropathy. Neurology, 2016, 86, 1762-1771.	1.1	52

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109	Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 846-863.	1.9	51
110	The Cardiac Genome Clinic: implementing genome sequencing in pediatric heart disease. Genetics in Medicine, 2020, 22, 1015-1024.	2.4	51
111	New insights into the regulatory function of CYFIP1 in the context of WAVE- and FMRP-containing complexes. DMM Disease Models and Mechanisms, 2017, 10, 463-474.	2.4	49
112	Mutations in RAB39B in individuals with intellectual disability, autism spectrum disorder, and macrocephaly. Molecular Autism, 2017, 8, 59.	4.9	49
113	Complex Copy Number Variation of AMY1 does not Associate with Obesity in two East Asian Cohorts. Human Mutation, 2016, 37, 669-678.	2.5	48
114	Frequent appearance of novel protein-coding sequences by frameshift translation. Genomics, 2006, 88, 690-697.	2.9	47
115	Xâ€linked Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2012, 17, 9-13.	3.1	47
116	CAOSâ€”Episodic Cerebellar Ataxia, Areflexia, Optic Atrophy, and Sensorineural Hearing Loss. Journal of Child Neurology, 2015, 30, 1749-1756.	1.4	47
117	Rare Variants in MME, Encoding Metalloprotease Neprilysin, Are Linked to Late-Onset Autosomal-Dominant Axonal Polyneuropathies. American Journal of Human Genetics, 2016, 99, 607-623.	6.2	47
118	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	5.9	47
119	Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. Acta Neuropathologica Communications, 2015, 3, 44.	5.2	45
120	Whole-Genome Sequencing Suggests Schizophrenia Risk Mechanisms in Humans with 22q11.2 Deletion Syndrome. G3: Genes, Genomes, Genetics, 2015, 5, 2453-2461.	1.8	43
121	Internodal specializations of myelinated axons in the central nervous system. Cell and Tissue Research, 2001, 305, 53-66.	2.9	42
122	Loss of function of the cytoplasmic isoform of the protein laforin (EPM2A) causes Lafora progressive myoclonus epilepsy. Human Mutation, 2004, 23, 170-176.	2.5	42
123	Analysis of the human Sonic Hedgehog coding and promoter regions in sacral agenesis, triphalangeal thumb, and mirror polydactyly. Human Genetics, 1998, 102, 387-392.	3.8	41
124	Microdeletions ofELP4Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. Human Mutation, 2015, 36, 842-850.	2.5	41
125	Genome sequencing as a platform for pharmacogenetic genotyping: a pediatric cohort study. Npj Genomic Medicine, 2017, 2, 19.	3.8	41
126	Germline and somatic mutations in STXBP1 with diverse neurodevelopmental phenotypes. Neurology: Genetics, 2017, 3, e199.	1.9	41

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127	Axons modulate the expression of proteolipid protein in the CNS. Journal of Neuroscience Research, 1992, 32, 138-148.	2.9	40
128	Copy-number variations are enriched for neurodevelopmental genes in children with developmental coordination disorder. Journal of Medical Genetics, 2016, 53, 812-819.	3.2	40
129	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	1.2	40
130	Prevalence and orthopedic management of foot and ankle deformities in Charcotâ€“Marieâ€“Tooth disease. Muscle and Nerve, 2018, 57, 255-259.	2.2	39
131	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	7.6	39
132	X-linked vacuolar myopathies: Two separate loci and refined genetic mapping. Annals of Neurology, 2000, 47, 666-669.	5.3	38
133	Intraneural <scp>GJB</scp>1 gene delivery improves nerve pathology in a model of <scp>X</scp>â€linked <scp>C</scp>harcotâ€“<scp>M</scp>arieâ€“<scp>T</scp>ooth disease. Annals of Neurology, 2015, 78, 303-316.	5.3	38
134	De novo PMP2mutations in families with type 1 Charcotâ€“Marieâ€“Tooth disease. Brain, 2016, 139, 1649-1656.	7.6	37
135	Oxytocin Receptor Polymorphisms are Differentially Associated with Social Abilities across Neurodevelopmental Disorders. Scientific Reports, 2017, 7, 11618.	3.3	36
136	Environmental exposures associated with elevated risk for autism spectrum disorder may augment the burden of deleterious de novo mutations among probands. Molecular Psychiatry, 2022, 27, 710-730.	7.9	36
137	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	3.3	35
138	ARHGEF9 disease. Neurology: Genetics, 2017, 3, e148.	1.9	35
139	A multicenter retrospective study of charcotâ€marieâ€tooth disease type 4B (CMT4B) associated with mutations in myotubularinâ€related proteins (MTMRs). Annals of Neurology, 2019, 86, 55-67.	5.3	35
140	Characterization of the human myeloid leukemia-derived cell line GF-D8 by multiplex fluorescence in situ hybridization, subtelomeric probes, and comparative genomic hybridization. , 1999, 24, 213-221.		34
141	A novel AARS mutation in a family with dominant myeloneuropathy. Neurology, 2015, 84, 2040-2047.	1.1	33
142	Variation in SIPA1L2 is correlated with phenotype modification in Charcotâ€“ Marieâ€“ Tooth disease type 1A. Annals of Neurology, 2019, 85, 316-330.	5.3	33
143	Maturation-dependent apoptotic cell death of oligodendrocytes in myelin-deficient rats. , 1998, 54, 623-634.		32
144	Caspr2 autoantibodies target multiple epitopes. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e127.	6.0	32

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145	Care and cost consequences of pediatric whole genome sequencing compared to chromosome microarray. European Journal of Human Genetics, 2017, 25, 1303-1312.	2.8	32
146	Impact of DNA source on genetic variant detection from human whole-genome sequencing data. Journal of Medical Genetics, 2019, 56, 809-817.	3.2	32
147	Control of Long-Term Synaptic Potentiation and Learning by Alternative Splicing of the NMDA Receptor Subunit GluN1. Cell Reports, 2019, 29, 4285-4294.e5.	6.4	32
148	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. Genetics in Medicine, 2019, 21, 1021-1026.	2.4	32
149	The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. Molecular Autism, 2014, 5, 34.	4.9	31
150	Variable phenotype expression in a family segregating microdeletions of the NRXN1 and MBD5 autism spectrum disorder susceptibility genes. Npj Genomic Medicine, 2017, 2, .	3.8	31
151	Chitayat-Hall and Schaaf-Yang syndromes:a common aetiology: expanding the phenotype of MAGEL2-related disorders. Journal of Medical Genetics, 2018, 55, 316-321.	3.2	31
152	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31
153	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
154	Development of a high-resolution Y-chromosome microarray for improved male infertility diagnosis. Fertility and Sterility, 2014, 101, 1079-1085.e3.	1.0	30
155	Prenatal growth restriction, retinal dystrophy, diabetes insipidus and white matter disease: expanding the spectrum of PRPS1-related disorders. European Journal of Human Genetics, 2015, 23, 310-316.	2.8	30
156	De novo exon 1 deletion of AUTS2 gene in a patient with autism spectrum disorder and developmental delay: A case report and a brief literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1381-1385.	1.2	30
157	Epileptic Encephalopathy Caused by Mutations in the Guanine Nucleotide Exchange Factor DENND5A. American Journal of Human Genetics, 2016, 99, 1359-1367.	6.2	30
158	Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. Genome Medicine, 2017, 9, 105.	8.2	30
159	Myelinated axons fail to develop properly in a genetically authentic mouse model of Charcot-Marie-Tooth disease type 2E. Experimental Neurology, 2018, 308, 13-25.	4.1	30
160	Schwann cellâ€“derived periostin promotes autoimmune peripheral polyneuropathy via macrophage recruitment. Journal of Clinical Investigation, 2018, 128, 4727-4741.	8.2	30
161	Structural organization and chromosomal localization of the human Na,K-ATPase Î²3 subunit gene and pseudogene. Mammalian Genome, 1998, 9, 136-143.	2.2	29
162	Expanding the neurodevelopmental phenotypes of individuals with de novo KMT2A variants. Npj Genomic Medicine, 2019, 4, 9.	3.8	29

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163	Subgroup-specific alternative splicing in medulloblastoma. Acta Neuropathologica, 2012, 123, 485-499.	7.7	28
164	DIXDC1 Phosphorylation and Control of Dendritic Morphology Are Impaired by Rare Genetic Variants. Cell Reports, 2016, 17, 1892-1904.	6.4	28
165	Shared genetic risk between eating disorderâ€•and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880.	2.6	28
166	Copy Number Variation Analysis of 100 Twin Pairs Enriched for Neurodevelopmental Disorders. Twin Research and Human Genetics, 2018, 21, 1-11.	0.6	27
167	Communicating complex genomic information: A counselling approach derived from research experience with Autism Spectrum Disorder. Patient Education and Counseling, 2018, 101, 352-361.	2.2	27
168	Microduplications at the pseudoautosomalSHOXlocus in autism spectrum disorders and related neurodevelopmental conditions. Journal of Medical Genetics, 2016, 53, 536-547.	3.2	26
169	Genome-wide rare copy number variations contribute to genetic risk for transposition of the great arteries. International Journal of Cardiology, 2016, 204, 115-121.	1.7	26
170	Heterogeneity in clinical sequencing tests marketed for autism spectrum disorders. Npj Genomic Medicine, 2018, 3, 27.	3.8	26
171	DNA Methylation of the Oxytocin Receptor Across Neurodevelopmental Disorders. Journal of Autism and Developmental Disorders, 2021, 51, 3610-3623.	2.7	26
172	Holoprosencephaly, sacral anomalies, and situs ambiguus in an infant with partial monosomy 7q/trisomy 2p and SHH and HLXB9 haploinsufficiency. Clinical Genetics, 2000, 57, 388-393.	2.0	25
173	Rare de novo deletion of metabotropic glutamate receptor 7 (GRM7) gene in a patient with autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2015, 168, 258-264.	1.7	25
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