Stephen W Scherer

List of Publications by Year
in descending order

308 papers	31,946 citations	10351 72 h-index	5227 165 g-index
318 all docs	318 docs citations	318 times ranked	41845 citing authors

#	Article	IF	Citations
1	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.	4.1	36
2	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	3.7	69
3	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	0.7	114
4	Copy number variations in a Brazilian cohort with autism spectrum disorders highlight the contribution of cell adhesion genes. Clinical Genetics, 2022, 101, 134-141.	1.0	13
5	A Regional Burden of Sequence-Level Variation in the 22q11.2 Region Influences Schizophrenia Risk and Educational Attainment. Biological Psychiatry, 2022, 91, 718-726.	0.7	1
6	Deletion of Loss-of-Functionâ€“Intolerant Genes and Risk of 5 Psychiatric Disorders. JAMA Psychiatry, 2022, 79, 78.	6.0	8
7	Mutational Landscape of Autism Spectrum Disorder Brain Tissue. Genes, 2022, 13, 207.	1.0	7
8	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	1.1	5
9	Chromosomal microarray analysis of 410 Han Chinese patients with autism spectrum disorder or unexplained intellectual disability and developmental delay. Npj Genomic Medicine, 2022, 7, 1.	1.7	11
10	GeneTerpret: a customizable multilayer approach to genomic variant prioritization and interpretation. BMC Medical Genomics, 2022, 15, 31.	0.7	1
11	Analysis of recent shared ancestry in a familial cohort identifies coding and noncoding autism spectrum disorder variants. Npj Genomic Medicine, 2022, 7, 13.	1.7	18
12	Complex Autism Spectrum Disorder with Epilepsy, Strabismus and Self-Injurious Behaviors in a Patient with a De Novo Heterozygous POLR2A Variant. Genes, 2022, 13, 470.	1.0	3
13	Biallelic PAN2 variants in individuals with a syndromic neurodevelopmental disorder and multiple congenital anomalies. European Journal of Human Genetics, 2022, 30, 611-618.	1.4	4
14	Whole genome sequencing delineates regulatory, copy number, and cryptic splice variants in early onset cardiomyopathy. Npj Genomic Medicine, 2022, 7, 18.	1.7	14
15	DNA methylation signature associated with Bohring-Opitz syndrome: a new tool for functional classification of variants in ASXL genes. European Journal of Human Genetics, 2022, 30, 695-702.	1.4	15
16	Genome-wide tandem repeat expansions contribute to schizophrenia risk. Molecular Psychiatry, 2022, 27, 3692-3698.	4.1	20
17	Mutations in trpÎ³, the homologue of TRPC6 autism candidate gene, causes autism-like behavioral deficits in Drosophila. Molecular Psychiatry, 2022, 27, 3328-3342.	4.1	6
18	Disruption of endosomal sorting in Schwann cells leads to defective myelination and endosomal abnormalities observed in Charcot-Marie-Tooth disease. Journal of Neuroscience, 2022, , JN-RM-2481-21.	1.7	1

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19	Germline PTPN13 mutations in patients with bone marrow failure and acute lymphoblastic leukemia. Leukemia, 2022, 36, 2132-2135.	3.3	1
20	Chromosomal-level reference genome assembly of the North American wolverine (Gulo gulo) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 70	0.8	2
21	Shared genetic risk between eating disorderâ€•and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880.	1.4	28
22	Adaptation and validation of the Genetic Counseling Outcome Scale for autism spectrum disorders and related conditions. Journal of Genetic Counseling, 2021, 30, 305-318.	0.9	7
23	DNA Methylation of the Oxytocin Receptor Across Neurodevelopmental Disorders. Journal of Autism and Developmental Disorders, 2021, 51, 3610-3623.	1.7	26
24	Long-Read Sequencing Improves the Detection of Structural Variations Impacting Complex Non-Coding Elements of the Genome. International Journal of Molecular Sciences, 2021, 22, 2060.	1.8	14
25	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. Molecular Psychiatry, 2021, 26, 1706-1718.	4.1	10
26	Whole exome sequencing uncovered highly penetrant recessive mutations for a spectrum of rare genetic pediatric diseases in Bangladesh. Npj Genomic Medicine, 2021, 6, 14.	1.7	8
27	Enrichment of loss-of-function and copy number variants in ventricular cardiomyopathy genes in â€˜loneâ€™ atrial fibrillation. Europace, 2021, 23, 844-850.	0.7	15
28	A recurrent MORC2 mutation causes Charcotâ€Marieâ€Tooth disease type 2Z. Journal of the Peripheral Nervous System, 2021, 26, 184-186.	1.4	3
29	Inducible knockout of Clec16a in mice results in sensory neurodegeneration. Scientific Reports, 2021, 11, 9319.	1.6	7
30	An Epigenetically Distinct Subset of Children With Autism Spectrum Disorder Resulting From Differences in Blood Cell Composition. Frontiers in Neurology, 2021, 12, 612817.	1.1	5
31	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.	2.6	31
32	What a finding of gene copy number variation can add to the diagnosis of developmental neuropsychiatric disorders. Current Opinion in Genetics and Development, 2021, 68, 18-25.	1.5	15
33	Temporal trends and yield of clinical diagnostic genetic testing in adult neurology. American Journal of Medical Genetics, Part A, 2021, 185, 2922-2928.	0.7	12
34	Genome sequencing for detection of pathogenic deep intronic variation: A clinical case report illustrating opportunities and challenges. American Journal of Medical Genetics, Part A, 2021, 185, 3129-3135.	0.7	10
35	Contribution of Multiple Inherited Variants to Autism Spectrum Disorder (ASD) in a Family with 3 Affected Siblings. Genes, 2021, 12, 1053.	1.0	12
36	Discovery of genomic variation across a generation. Human Molecular Genetics, 2021, 30, R174-R186.	1.4	9

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37	Clinical Genetic Risk Variants Inform a Functional Protein Interaction Network for Tetralogy of Fallot. Circulation Genomic and Precision Medicine, 2021, 14, e003410.	1.6	15
38	Sleep phenotype of individuals with autism spectrum disorder bearing mutations in the <scp>PER2</scp> circadian rhythm gene. American Journal of Medical Genetics, Part A, 2021, 185, 1120-1130.	0.7	17
39	Genome sequencing as a diagnostic test. Cmaj, 2021, 193, E1626-E1629.	0.9	20
40	Axonal Charcot-Marie-Tooth Disease: from Common Pathogenic Mechanisms to Emerging Treatment Opportunities. Neurotherapeutics, 2021, 18, 2269-2285.	2.1	25
41	Predictors of empowerment in parents of children with autism and related neurodevelopmental disorders who are undergoing genetic testing. Molecular Genetics & Genomic Medicine, 2021, 9, e1803.	0.6	2
42	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91.	1.7	9
43	Homozygous duplication identified by whole genome sequencing causes LRBA deficiency. Npj Genomic Medicine, 2021, 6, 96.	1.7	3
44	Single-cell transcriptome identifies molecular subtype of autism spectrum disorder impacted by de novo loss-of-function variants regulating glial cells. Human Genomics, 2021, 15, 68.	1.4	20
45	VikNGS: a C++ variant integration kit for next generation sequencing association analysis. Bioinformatics, 2020, 36, 1283-1285.	1.8	10
46	Synaptic Dysfunction in Human Neurons With Autism-Associated Deletions in PTCHD1-AS. Biological Psychiatry, 2020, 87, 139-149.	0.7	57
47	Genes and Pathways Implicated in Tetralogy of Fallot Revealed by Ultra-Rare Variant Burden Analysis in 231 Genome Sequences. Frontiers in Genetics, 2020, 11, 957.	1.1	23
48	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	2.8	47
49	Isoform-specific loss of dystonin causes hereditary motor and sensory neuropathy. Neurology: Genetics, 2020, 6, e496.	0.9	9
50	Reliability of the <scp>Charcotâ€Marieâ€Tooth</scp> functional outcome measure. Journal of the Peripheral Nervous System, 2020, 25, 288-291.	1.4	8
51	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	13.7	155
52	A Distributed Whole Genome Sequencing Benchmark Study. Frontiers in Genetics, 2020, 11, 612515.	1.1	6
53	Phase Separation as a Missing Mechanism for Interpretation of Disease Mutations. Cell, 2020, 183, 1742-1756.	13.5	147
54	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1912-1918.	15.2	90

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55	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	9.4	97
56	Modeling neuronal consequences of autism-associated gene regulatory variants with human induced pluripotent stem cells. Molecular Autism, 2020, 11, 33.	2.6	6
57	Ancestry and frequency of genetic variants in the general population are confounders in the characterization of germline variants linked to cancer. BMC Medical Genetics, 2020, 21, 92.	2.1	4
58	Segregating patterns of copy number variations in extended autism spectrum disorder (ASD) pedigrees. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 268-276.	1.1	7
59	A recessive Trim2 mutation causes an axonal neuropathy in mice. Neurobiology of Disease, 2020, 140, 104845.	2.1	12
60	Perceived utility of biological testing for autism spectrum disorder is associated with child and family functioning. Research in Developmental Disabilities, 2020, 100, 103605.	1.2	7
61	The Cardiac Genome Clinic: implementing genome sequencing in pediatric heart disease. Genetics in Medicine, 2020, 22, 1015-1024.	1.1	51
62	Refining critical regions in 15q24 microdeletion syndrome pertaining to autism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 217-226.	1.1	2
63	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
64	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	2.6	59
65	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	7.7	83
66	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
67	TUBB4A mutations result in both glial and neuronal degeneration in an H-ABC leukodystrophy mouse model. ELife, 2020, 9, .	2.8	15
68	Haploinsufficiency in the ANKS1B gene encoding AIDA-1 leads to a neurodevelopmental syndrome. Nature Communications, 2019, 10, 3529.	5.8	20
69	A MT-ATP6 Mutation Causes a Slowly Progressive Myeloneuropathy. Journal of Neuromuscular Diseases, 2019, 6, 385-387.	1.1	2
70	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	0.7	25
71	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
72	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	3.7	31

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73	Genome-wide copy number variant data for inflammatory bowel disease in a caucasian population. Data in Brief, 2019, 25, 104203.	0.5	0
74	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	1.7	118
75	A recurrent GARS mutation causes distal hereditary motor neuropathy. Journal of the Peripheral Nervous System, 2019, 24, 320-323.	1.4	12
76	Yield of nextâ€generation neuropathy gene panels in axonal neuropathies. Journal of the Peripheral Nervous System, 2019, 24, 324-329.	1.4	7
77	Impact of DNA source on genetic variant detection from human whole-genome sequencing data. Journal of Medical Genetics, 2019, 56, 809-817.	1.5	32
78	Association between distress and knowledge among parents of autistic children. PLoS ONE, 2019, 14, e0223119.	1.1	5
79	Variation in SIPA1L2 is correlated with phenotype modification in Charcotâ€“ Marieâ€“ Tooth disease type 1A. Annals of Neurology, 2019, 85, 316-330.	2.8	33
80	STXBP1 encephalopathy is associated with awake bruxism. Epilepsy and Behavior, 2019, 92, 121-124.	0.9	18
81	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
82	Genome-wide analysis identifies rare copy number variations associated with inflammatory bowel disease. PLoS ONE, 2019, 14, e0217846.	1.1	16
83	Expanding the neurodevelopmental phenotypes of individuals with de novo KMT2A variants. Npj Genomic Medicine, 2019, 4, 9.	1.7	29
84	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.	2.8	35
85	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
86	Modifier Gene Candidates in Charcot-Marie-Tooth Disease Type 1A: A Case-Only Genome-Wide Association Study. Journal of Neuromuscular Diseases, 2019, 6, 201-211.	1.1	19
87	POLG mutations presenting as Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2019, 24, 213-218.	1.4	6
88	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.	2.4	63
89	A Third Linear Association Between Olduvai (DUF1220) Copy Number and Severity of the Classic Symptoms of Inherited Autism. American Journal of Psychiatry, 2019, 176, 643-650.	4.0	16
90	Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3.	1.5	6

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91	Reanalysing genomic data by normalized coverage values uncovers CNVs in bone marrow failure gene panels. Npj Genomic Medicine, 2019, 4, 30.	1.7	3
92	Critical exon indexing improves clinical interpretation of copy number variants in neurodevelopmental disorders. Neurology: Genetics, 2019, 5, e378.	0.9	4
93	Analysis of five deep-sequenced trio-genomes of the Peninsular Malaysia Orang Asli and North Borneo populations. BMC Genomics, 2019, 20, 842.	1.2	3
94	Control of Long-Term Synaptic Potentiation and Learning by Alternative Splicing of the NMDA Receptor Subunit GluN1. Cell Reports, 2019, 29, 4285-4294.e5.	2.9	32
95	Precision Health Resource of Control iPSC Lines for Versatile Multilineage Differentiation. Stem Cell Reports, 2019, 13, 1126-1141.	2.3	24
96	A novel MFN2 mutation causes variable clinical severity in a multi-generational CMT2 family. Neuromuscular Disorders, 2019, 29, 134-137.	0.3	5
97	Rare copy number variation in extremely impulsively violent males. Genes, Brain and Behavior, 2019, 18, e12536.	1.1	9
98	Biallelic mutations in EXOC3L2 cause a novel syndrome that affects the brain, kidney and blood. Journal of Medical Genetics, 2019, 56, 340-346.	1.5	9
99	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. Genetics in Medicine, 2019, 21, 1021-1026.	1.1	32
100	Haploinsufficiency of vascular endothelial growth factor related signaling genes is associated with tetralogy of Fallot. Genetics in Medicine, 2019, 21, 1001-1007.	1.1	58
101	Altered TAOK2 activity causes autism-related neurodevelopmental and cognitive abnormalities through RhoA signaling. Molecular Psychiatry, 2019, 24, 1329-1350.	4.1	128
102	CNTN5-/+or EHMT2-/+human iPSC-derived neurons from individuals with autism develop hyperactive neuronal networks. ELife, 2019, 8, .	2.8	72
103	Variant pathogenicity evaluation in the community-driven Inherited Neuropathy Variant Browser. Human Mutation, 2018, 39, 635-642.	1.1	13
104	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	2.6	59
105	Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331.	6.0	174
106	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136.	0.9	57
107	Progress in the genetics of autism spectrum disorder. Developmental Medicine and Child Neurology, 2018, 60, 445-451.	1.1	116
108	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.	1.4	88

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109	OTUD7A Regulates Neurodevelopmental Phenotypes in the 15q13.3 Microdeletion Syndrome. American Journal of Human Genetics, 2018, 102, 278-295.	2.6	81
110	A mutation in the heptad repeat 2 domain of MFN2 in a large CMT2A family. Journal of the Peripheral Nervous System, 2018, 23, 36-39.	1.4	5
111	Copy Number Variation Analysis of 100 Twin Pairs Enriched for Neurodevelopmental Disorders. Twin Research and Human Genetics, 2018, 21, 1-11.	0.3	27
112	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
113	Chitayat-Hall and Schaaf-Yang syndromes:a common aetiology: expanding the phenotype of MAGEL2-related disorders. Journal of Medical Genetics, 2018, 55, 316-321.	1.5	31
114	Copy number variation in fetal alcohol spectrum disorder. Biochemistry and Cell Biology, 2018, 96, 161-166.	0.9	15
115	Improving imputation in disease-relevant regions: lessons from cystic fibrosis. Npj Genomic Medicine, 2018, 3, 8.	1.7	9
116	Genetic test utilization and diagnostic yield in adult patients with neurological disorders. Neurogenetics, 2018, 19, 105-110.	0.7	20
117	Prevalence and orthopedic management of foot and ankle deformities in Charcotâ€“Marieâ€“Tooth disease. Muscle and Nerve, 2018, 57, 255-259.	1.0	39
118	Carpal tunnel syndrome in inherited neuropathies: A retrospective survey. Muscle and Nerve, 2018, 57, 388-394.	1.0	14
119	Neurofascin antibodies in autoimmune, genetic, and idiopathic neuropathies. Neurology, 2018, 90, e31-e38.	1.5	78
120	Association of IMMP2L deletions with autism spectrum disorder: A trio family study and metaâ€analysis. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 93-100.	1.1	16
121	De novo and rare inherited copy-number variations in the hemiplegic form of cerebral palsy. Genetics in Medicine, 2018, 20, 172-180.	1.1	82
122	Communicating complex genomic information: A counselling approach derived from research experience with Autism Spectrum Disorder. Patient Education and Counseling, 2018, 101, 352-361.	1.0	27
123	Nodes, paranodes and neuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 61-71.	0.9	60
124	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
125	Complete Disruption of Autism-Susceptibility Genes by Gene Editing Predominantly Reduces Functional Connectivity of Isogenic Human Neurons. Stem Cell Reports, 2018, 11, 1211-1225.	2.3	111
126	Myopathy associated BAG3 mutations lead to protein aggregation by stalling Hsp70 networks. Nature Communications, 2018, 9, 5342.	5.8	65

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127	Heterogeneity in clinical sequencing tests marketed for autism spectrum disorders. Npj Genomic Medicine, 2018, 3, 27.	1.7	26
128	Rearrangement bursts generate canonical gene fusions in bone and soft tissue tumors. Science, 2018, 361, .	6.0	121
129	Chorea-acanthocytosis. Neurology: Genetics, 2018, 4, e242.	0.9	4
130	Genome-wide copy number variation analysis identifies novel candidate loci associated with pediatric obesity. European Journal of Human Genetics, 2018, 26, 1588-1596.	1.4	23
131	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.	2.0	30
132	A genome-wide linkage study of autism spectrum disorder and the broad autism phenotype in extended pedigrees. Journal of Neurodevelopmental Disorders, 2018, 10, 20.	1.5	20
133	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
134	Paternal uniparental disomy of chromosome 19 in a pair of monochorionic diamniotic twins with dysmorphic features and developmental delay. Journal of Medical Genetics, 2018, 55, 847-852.	1.5	6
135	Schwann cellâ€“derived periostin promotes autoimmune peripheral polyneuropathy via macrophage recruitment. Journal of Clinical Investigation, 2018, 128, 4727-4741.	3.9	30
136	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.	1.1	70
137	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
138	Biallelic mutations in DNAJC21 cause Shwachman-Diamond syndrome. Blood, 2017, 129, 1557-1562.	0.6	104
139	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.	1.2	49
140	De NovoGenome and Transcriptome Assembly of the Canadian Beaver (Castor canadensis). G3: Genes, Genomes, Genetics, 2017, 7, 755-773.	0.8	18
141	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	7.1	691
142	A de novo deletion in a boy with cerebral palsy suggests a refined critical region for the 4q21.22 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1287-1293.	0.7	8
143	Variable phenotype expression in a family segregating microdeletions of the NRXN1 and MBD5 autism spectrum disorder susceptibility genes. Npj Genomic Medicine, 2017, 2, .	1.7	31
144	Copy Number Variation in Tourette Syndrome. Neuron, 2017, 94, 1041-1043.	3.8	6

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145	Febrile ataxia and myokymia broaden the SPG26 hereditary spastic paraplegia phenotype. Neurology: Genetics, 2017, 3, e156.	0.9	7
146	ARHGEF9 disease. Neurology: Genetics, 2017, 3, e148.	0.9	35
147	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
148	Hyperventilation-athetosis in ASXL3 deficiency (Bainbridge-Ropers) syndrome. Neurology: Genetics, 2017, 3, e189.	0.9	7
149	De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. , 2017, 173, 2725-2730.		15
150	Oxytocin Receptor Polymorphisms are Differentially Associated with Social Abilities across Neurodevelopmental Disorders. Scientific Reports, 2017, 7, 11618.	1.6	36
151	HLX is a candidate gene for a pattern of anomalies associated with congenital diaphragmatic hernia, short bowel, and asplenia. American Journal of Medical Genetics, Part A, 2017, 173, 3070-3074.	0.7	10
152	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. Nature Neuroscience, 2017, 20, 1217-1224.	7.1	212
153	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	4.0	77
154	Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 846-863.	0.9	51
155	Care and cost consequences of pediatric whole genome sequencing compared to chromosome microarray. European Journal of Human Genetics, 2017, 25, 1303-1312.	1.4	32
156	Genome sequencing as a platform for pharmacogenetic genotyping: a pediatric cohort study. Npj Genomic Medicine, 2017, 2, 19.	1.7	41
157	The clinical impact of copy number variants in inherited bone marrow failure syndromes. Npj Genomic Medicine, 2017, 2, .	1.7	10
158	Use of clinical chromosomal microarray in Chinese patients with autism spectrum disorderâ€"implications of a copy number variation involving DPP10. Molecular Autism, 2017, 8, 31.	2.6	16
159	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	0.7	40
160	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
161	Atypical autism in a boy with double duplication of 22q11.2: implications of increasing dosage. Npj Genomic Medicine, 2017, 2, 28.	1.7	3
162	Characterization of Large Copy Number Variation in Mexican Type 2 Diabetes subjects. Scientific Reports, 2017, 7, 17105.	1.6	10

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163	Germline and somatic mutations in STXBP1 with diverse neurodevelopmental phenotypes. Neurology: Genetics, 2017, 3, e199.	0.9	41
164	Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. Genome Medicine, 2017, 9, 105.	3.6	30
165	Mutations in RAB39B in individuals with intellectual disability, autism spectrum disorder, and macrocephaly. Molecular Autism, 2017, 8, 59.	2.6	49
166	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
167	A microcosting and costâ€“consequence analysis of clinical genomic testing strategies in autism spectrum disorder. Genetics in Medicine, 2017, 19, 1268-1275.	1.1	62
168	PMP22 antisense oligonucleotides reverse Charcot-Marie-Tooth disease type 1A features in rodent models. Journal of Clinical Investigation, 2017, 128, 359-368.	3.9	117
169	Whole-genome sequencing suggests mechanisms for 22q11.2 deletion-associated Parkinsonâ€™s disease. PLoS ONE, 2017, 12, e0173944.	1.1	17
170	Syndromic autism spectrum disorders: moving from a clinically defined to a molecularly defined approach. Dialogues in Clinical Neuroscience, 2017, 19, 353-371.	1.8	105
171	MED23â€associated refractory epilepsy successfully treated with the ketogenic diet. American Journal of Medical Genetics, Part A, 2016, 170, 2421-2425.	0.7	21
172	The 22q11 PRODH/DGCR6 deletion is frequent in hyperprolinemic subjects but is not a strong risk factor for ASD. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 377-382.	1.1	3
173	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	1.7	295
174	Copy-number variations are enriched for neurodevelopmental genes in children with developmental coordination disorder. Journal of Medical Genetics, 2016, 53, 812-819.	1.5	40
175	De novo large rare copy-number variations contribute to conotruncal heart disease in Chinese patients. Npj Genomic Medicine, 2016, 1, 16033.	1.7	8
176	Target-enrichment sequencing and copy number evaluation in inherited polyneuropathy. Neurology, 2016, 86, 1762-1771.	1.5	52
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