Christian R Marshall

List of Publications by Year
in descending order

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135 papers	14,996 citations	44069 48 h-index	20961 115 g-index
144 all docs	144 docs citations	144 times ranked	20265 citing authors

#	Article	IF	Citations
1	Within-family influences on dimensional neurobehavioral traits in a high-risk genetic model. Psychological Medicine, 2022, 52, 3184-3192.	4.5	11
2	Data sharing to improve concordance in variant interpretation across laboratories: results from the Canadian Open Genetics Repository. Journal of Medical Genetics, 2022, 59, 571-578.	3.2	14
3	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	1.3	114
4	Genome sequencing among children with medical complexity: What constitutes value from parentsâ€™ perspective?. Journal of Genetic Counseling, 2022, 31, 523-533.	1.6	5
5	Hereditary Mucin Deficiency Caused by Biallelic Loss of Function of MUC5B. American Journal of Respiratory and Critical Care Medicine, 2022, 205, 761-768.	5.6	12
6	Diagnostic yield of genome sequencing for prenatal diagnosis of fetal structural anomalies. Prenatal Diagnosis, 2022, 42, 822-830.	2.3	12
7	Trio genome sequencing for developmental delay and pediatric heart conditions: A comparative microcost analysis. Genetics in Medicine, 2022, 24, 1027-1036.	2.4	7
8	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. Human Mutation, 2022, , .	2.5	4
9	A Chromosomal Duplication Encompassing Interleukin-33 Causes a Novel Hyper IgE Phenotype Characterized by Eosinophilic Esophagitis and Generalized Autoimmunity. Gastroenterology, 2022, 163, 510-513.e3.	1.3	8
10	Genome-wide tandem repeat expansions contribute to schizophrenia risk. Molecular Psychiatry, 2022, 27, 3692-3698.	7.9	20
11	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
12	Whole genome sequencing reveals biallelic <scp>PLA2G6</scp> mutations in siblings with cerebellar atrophy and cap myopathy. Clinical Genetics, 2021, 99, 746-748.	2.0	3
13	Genome sequencing broadens the range of contributing variants with clinical implications in schizophrenia. Translational Psychiatry, 2021, 11, 84.	4.8	16
14	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.	1.2	10
15	Mild Idiopathic Infantile Hypercalcemiaâ€”Part 1: Biochemical and Genetic Findings. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 2915-2937.	3.6	8
16	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679.	3.5	17
17	Clinical Genetic Risk Variants Inform a Functional Protein Interaction Network for Tetralogy of Fallot. Circulation Genomic and Precision Medicine, 2021, 14, e003410.	3.6	15
18	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	7.9	87

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19	Genome sequencing as a diagnostic test. Cmaj, 2021, 193, E1626-E1629.	2.0	20
20	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91.	3.8	9
21	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.	2.3	23
22	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	5.9	47
23	A novel intronic variant in UBE3A identified by genome sequencing in a patient with an atypical presentation of Angelman syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2145-2151.	1.2	3
24	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47.	3.8	67
25	Genome sequencing identifies a rare case of moderate Zellweger spectrum disorder caused by a PEX3 defect: Case report and literature review. Molecular Genetics and Metabolism Reports, 2020, 25, 100664.	1.1	1
26	The Medical Genome Initiative: moving whole-genome sequencing for rare disease diagnosis to the clinic. Genome Medicine, 2020, 12, 48.	8.2	40
27	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. Frontiers in Molecular Neuroscience, 2020, 13, 12.	2.9	12
28	The Cardiac Genome Clinic: implementing genome sequencing in pediatric heart disease. Genetics in Medicine, 2020, 22, 1015-1024.	2.4	51
29	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
30	Thiemann disease and familial digital arthropathy â€“ brachydactyly: two sides of the same coin?. Orphanet Journal of Rare Diseases, 2019, 14, 156.	2.7	3
31	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
32	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	3.8	118
33	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
34	Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3.	3.1	6
35	Analysis of five deep-sequenced trio-genomes of the Peninsular Malaysia Orang Asli and North Borneo populations. BMC Genomics, 2019, 20, 842.	2.8	3
36	Bi-allelic mutations ofLONP1encoding the mitochondrial LonP1 protease cause pyruvate dehydrogenase deficiency and profound neurodegeneration with progressive cerebellar atrophy. Human Molecular Genetics, 2019, 28, 290-306.	2.9	27

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37	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. Genetics in Medicine, 2019, 21, 1021-1026.	2.4	32
38	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
39	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136.	2.0	57
40	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
41	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163.	2.9	22
42	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
43	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
44	Copy number variation in fetal alcohol spectrum disorder. Biochemistry and Cell Biology, 2018, 96, 161-166.	2.0	15
45	Prospective cohort study for identification of underlying genetic causes in neonatal encephalopathy using whole-exome sequencing. Genetics in Medicine, 2018, 20, 486-494.	2.4	38
46	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
47	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
48	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.	3.2	6
49	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
50	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
51	Congenital myopathy with â€œcoronaâ€•fibres, selective muscle atrophy, and craniosynostosis associated with novel recessive mutations in SCN4A. Neuromuscular Disorders, 2017, 27, 574-580.	0.6	23
52	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	14.8	691
53	ARHGEF9 disease. Neurology: Genetics, 2017, 3, e148.	1.9	35
54	Severe neurodegeneration, progressive cerebral volume loss and diffuse hypomyelination associated with a homozygous frameshift mutation in CSTB. European Journal of Human Genetics, 2017, 25, 775-778.	2.8	24

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55	De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. , 2017, 173, 2725-2730.		15
56	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.	1.2	10
57	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
58	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
59	Genome sequencing as a platform for pharmacogenetic genotyping: a pediatric cohort study. Npj Genomic Medicine, 2017, 2, 19.	3.8	41
60	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.	4.9	16
61	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	1.2	40
62	Neuropsychiatric aspects of 22q11.2 deletion syndrome: considerations in the prenatal setting. Prenatal Diagnosis, 2017, 37, 61-69.	2.3	13
63	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
64	Germline and somatic mutations in STXBP1 with diverse neurodevelopmental phenotypes. Neurology: Genetics, 2017, 3, e199.	1.9	41
65	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression. , 2017, 58, 1736.		17
66	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
67	Mutations in RAB39B in individuals with intellectual disability, autism spectrum disorder, and macrocephaly. Molecular Autism, 2017, 8, 59.	4.9	49
68	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
69	Whole-genome sequencing suggests mechanisms for 22q11.2 deletion-associated Parkinsonâ€™s disease. PLoS ONE, 2017, 12, e0173944.	2.5	17
70	MED23â€associated refractory epilepsy successfully treated with the ketogenic diet. American Journal of Medical Genetics, Part A, 2016, 170, 2421-2425.	1.2	21
71	Arginine-Glycine Amidinotransferase Deficiency and Functional Characterization of Missense Variants inGATM. Human Mutation, 2016, 37, 926-932.	2.5	4
72	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295

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73	De novo large rare copy-number variations contribute to conotruncal heart disease in Chinese patients. Npj Genomic Medicine, 2016, 1, 16033.	3.8	8
74	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
75	Microcephalyâ€capillary malformation syndrome: Brothers with a homozygous STAMBP mutation, uncovered by exome sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 3018-3022.	1.2	16
76	Epileptic Encephalopathy Caused by Mutations in the Guanine Nucleotide Exchange Factor DENND5A. American Journal of Human Genetics, 2016, 99, 1359-1367.	6.2	30
77	Genome-wide characteristics of de novo mutations in autism. Npj Genomic Medicine, 2016, 1, 160271-1602710.	3.8	200
78	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
79	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	3.3	35
80	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
81	A recurrent germline mutation in the PIGA gene causes Simpsonâ€Golabiâ€Behmel syndrome type 2. American Journal of Medical Genetics, Part A, 2016, 170, 392-402.	1.2	34
82	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
83	Clinical delineation of the PACS1â€related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	1.2	44
84	Lethal Disorder of Mitochondrial Fission Caused by Mutations in DNM1L. Journal of Pediatrics, 2016, 171, 313-316.e2.	1.8	67
85	Rare Copy Number Variants Identified Suggest the Regulating Pathways in Hypertension-Related Left Ventricular Hypertrophy. PLoS ONE, 2016, 11, e0148755.	2.5	8
86	15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693.	2.5	67
87	MG-132â€...Diagnostic utility of whole genome sequencing in paediatric medicine. Journal of Medical Genetics, 2015, 52, A12.1-A12.	3.2	1
88	MG-108â€...Beyond the ACMG 56: Parental choices and initial results from a comprehensive whole genome sequencing-based search for predictive genomic variants in children. Journal of Medical Genetics, 2015, 52, A3.2-A4.	3.2	0
89	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
90	MG-129â€...Our experience ofin silicogene panel testing for clinically heterogeneous disorders using exome sequencing. Journal of Medical Genetics, 2015, 52, A11.1-A11.	3.2	1

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91	MG-130â€…Utilising whole exome sequencing to identify causative variants in genetically heterogeneous disorders. Journal of Medical Genetics, 2015, 52, A11.2-A11.	3.2	0
92	MG-106â€…Global developmental delay and characteristic facial features associated with pacs1 gene mutation â€“ report of two cases. Journal of Medical Genetics, 2015, 52, A1.2-A1.	3.2	0
93	Microdeletions ofELP4Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. Human Mutation, 2015, 36, 842-850.	2.5	41
94	MG-123â€…Exonic and intronic NRXN1 deletions: Novel genotype-phenotype correlations. Journal of Medical Genetics, 2015, 52, A9.1-A9.	3.2	0
95	Estimated carrier frequency of creatine transporter deficiency in females in the general population using functional characterization of novel missense variants in the SLC6A8 gene. Gene, 2015, 565, 187-191.	2.2	25
96	Carrier frequency of guanidinoacetate methyltransferase deficiency in the general population by functional characterization of missense variants in the GAMT gene. Molecular Genetics and Genomics, 2015, 290, 2163-2171.	2.1	23
97	Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847.	6.2	22
98	MG-108â€…Agenesis of the corpus callosum and autism associated with zeb1 gene deletion â€“ a case report. Journal of Medical Genetics, 2015, 52, A2.1-A2.	3.2	0
99	Whole-genome sequencing of quartet families with autism spectrum disorder. Nature Medicine, 2015, 21, 185-191.	30.7	457
100	A high-resolution copy-number variation resource for clinical and population genetics. Genetics in Medicine, 2015, 17, 747-752.	2.4	73
101	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
102	Whole-Exome Sequencing and Targeted Copy Number Analysis in Primary Ciliary Dyskinesia. G3: Genes, Genomes, Genetics, 2015, 5, 1775-1781.	1.8	53
103	CAOSâ€”Episodic Cerebellar Ataxia, Areflexia, Optic Atrophy, and Sensorineural Hearing Loss. Journal of Child Neurology, 2015, 30, 1749-1756.	1.4	47
104	PMPCAmutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517.	7.6	58
105	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
106	Clinically relevant copy number variations detected in cerebral palsy. Nature Communications, 2015, 6, 7949.	12.8	120
107	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
108	ISDN2014_0253: High resolution genomic analyses of a clinically defined autism spectrum disorder cohort. International Journal of Developmental Neuroscience, 2015, 47, 76-76.	1.6	2

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109	Identification of a novel MSH6 germline variant in a family with multiple gastro-intestinal malignancies by next generation sequencing. Familial Cancer, 2015, 14, 69-75.	1.9	1
110	Copy Number Variable MicroRNAs in Schizophrenia and Their Neurodevelopmental Gene Targets. Biological Psychiatry, 2015, 77, 158-166.	1.3	58
111	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
112	Novel Population Specific Autosomal Copy Number Variation and Its Functional Analysis amongst Negritos from Peninsular Malaysia. PLoS ONE, 2014, 9, e100371.	2.5	6
113	OTX2mutations cause autosomal dominant pattern dystrophy of the retinal pigment epithelium. Journal of Medical Genetics, 2014, 51, 797-805.	3.2	40
114	Complex genomic rearrangements in the dystrophin gene due to replicationâ€based mechanisms. Molecular Genetics & Genomic Medicine, 2014, 2, 539-547.	1.2	16
115	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	6.2	819
116	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
117	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.	2.9	140
118	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
119	CHD2 haploinsufficiency is associated with developmental delay, intellectual disability, epilepsy and neurobehavioural problems. Journal of Neurodevelopmental Disorders, 2014, 6, 9.	3.1	71
120	Recurrent duplications of the annexin A1 gene (ANXA1) in autism spectrum disorders. Molecular Autism, 2014, 5, 28.	4.9	13
121	Development of a high-resolution Y-chromosome microarray for improved male infertility diagnosis. Fertility and Sterility, 2014, 101, 1079-1085.e3.	1.0	30
122	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
123	Pathogenic rare copy number variants in community-based schizophrenia suggest a potential role for clinical microarrays. Human Molecular Genetics, 2013, 22, 4485-4501.	2.9	120
124	Detection of Clinically Relevant Genetic Variants in Autism Spectrum Disorder by Whole-Genome Sequencing. American Journal of Human Genetics, 2013, 93, 249-263.	6.2	429
125	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
126	Rare Copy Number Variations in Adults with Tetralogy of Fallot Implicate Novel Risk Gene Pathways. PLoS Genetics, 2012, 8, e1002843.	3.5	149

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127	Detection and Characterization of Copy Number Variation in Autism Spectrum Disorder. Methods in Molecular Biology, 2012, 838, 115-135.	0.9	72
128	SHANK1 Deletions in Males with Autism Spectrum Disorder. American Journal of Human Genetics, 2012, 90, 879-887.	6.2	292
129	Rare Copy Number Variation Discovery and Cross-Disorder Comparisons Identify Risk Genes for ADHD. Science Translational Medicine, 2011, 3, 95ra75.	12.4	304
130	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
131	Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68.	12.4	178
132	Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488.	6.2	1,641
133	Infantile Spasms Is Associated with Deletion of the MAGI2 Gene on Chromosome 7q11.23-q21.11. American Journal of Human Genetics, 2008, 83, 106-111.	6.2	108
134	Copy number variations and risk for schizophrenia in 22q11.2 deletion syndrome. Human Molecular Genetics, 2008, 17, 4045-4053.	2.9	155
135	Contribution of SHANK3 Mutations to Autism Spectrum Disorder. American Journal of Human Genetics, 2007, 81, 1289-1297.	6.2	604