Roger E Stevenson

List of Publications by Year
in descending order

93 papers	8,027 citations	81743 39 h-index	58464 82 g-index
94 all docs	94 docs citations	94 times ranked	10378 citing authors

#	Article	IF	Citations
1	Thermotolerance Generated by Plant/Fungal Symbiosis. Science, 2002, 298, 1581-1581.	6.0	682
2	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	9.4	528
3	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	9.4	509
4	Xâ€“linked spastic paraplegia (SPG1), MASA syndrome and Xâ€“linked hydrocephalus result from mutations in the L1 gene. Nature Genetics, 1994, 7, 402-407.	9.4	423
5	Evaluation of mental retardation: Recommendations of a consensus conference. , 1997, 72, 468-477.		344
6	Autism and maternally derived aberrations of chromosome 15q. , 1998, 76, 327-336.		324
7	Allan-Herndon-Dudley Syndrome and the Monocarboxylate Transporter 8 (MCT8) Gene. American Journal of Human Genetics, 2005, 77, 41-53.	2.6	314
8	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. Nature Genetics, 2007, 39, 1127-1133.	9.4	228
9	SLC9A6 Mutations Cause X-Linked Mental Retardation, Microcephaly, Epilepsy, and Ataxia, a Phenotype Mimicking Angelman Syndrome. American Journal of Human Genetics, 2008, 82, 1003-1010.	2.6	209
10	Fragile X and X-Linked Intellectual Disability: Four Decades of Discovery. American Journal of Human Genetics, 2012, 90, 579-590.	2.6	200
11	Mutations in CUL4B, Which Encodes a Ubiquitin E3 Ligase Subunit, Cause an X-linked Mental Retardation Syndrome Associated with Aggressive Outbursts, Seizures, Relative Macrocephaly, Central Obesity, Hypogonadism, Pes Cavus, and Tremor. American Journal of Human Genetics, 2007, 80, 345-352.	2.6	197
12	A recurrent mutation in MED12 leading to R961W causes Opitz-Kaveggia syndrome. Nature Genetics, 2007, 39, 451-453.	9.4	179
13	Recurrent Infections, Hypotonia, and Mental Retardation Caused by Duplication of MECP2 and Adjacent Region in Xq28. Pediatrics, 2006, 118, e1687-e1695.	1.0	173
14	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	2.6	171
15	A unique exonic splice enhancer mutation in a family with X-linked mental retardation and epilepsy points to a novel role of the renin receptor. Human Molecular Genetics, 2005, 14, 1019-1027.	1.4	167
16	Mutations in the planar cell polarity genesCELSR1andSCRIBare associated with the severe neural tube defect craniorachischisis. Human Mutation, 2012, 33, 440-447.	1.1	166
17	Mutations in the DLG3 Gene Cause Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 318-324.	2.6	157
18	The original Lujan syndrome family has a novel missense mutation (p.N1007S) in the MED12 gene. Journal of Medical Genetics, 2007, 44, 472-477.	1.5	153

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19	Autism and macrocephaly. Lancet, The, 1997, 349, 1744-1745.	6.3	137
20	A mixed epigenetic/genetic model for oligogenic inheritance of autism with a limited role forUBE3A. American Journal of Medical Genetics Part A, 2004, 131A, 1-10.	2.4	135
21	X-linked spermine synthase gene (SMS) defect: the first polyamine deficiency syndrome. European Journal of Human Genetics, 2003, 11, 937-944.	1.4	134
22	Mutations in the guanine nucleotide exchange factor gene IQSEC2 cause nonsyndromic intellectual disability. Nature Genetics, 2010, 42, 486-488.	9.4	134
23	Decreased tryptophan metabolism in patients with autism spectrum disorders. Molecular Autism, 2013, 4, 16.	2.6	124
24	Fine-Scale Survey of X Chromosome Copy Number Variants and Indels Underlying Intellectual Disability. American Journal of Human Genetics, 2010, 87, 173-188.	2.6	107
25	Mutations in ionotropic AMPA receptor 3 alter channel properties and are associated with moderate cognitive impairment in humans. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 18163-18168.	3.3	100
26	Xâ€linked intellectual disability update 2017. American Journal of Medical Genetics, Part A, 2018, 176, 1375-1388.	0.7	88
27	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. Genetics in Medicine, 2021, 23, 1065-1074.	1.1	88
28	Mutations in the BRWD3 Gene Cause X-Linked Mental Retardation Associated with Macrocephaly. American Journal of Human Genetics, 2007, 81, 367-374.	2.6	85
29	An X-linked channelopathy with cardiomegaly due to a CLIC2 mutation enhancing ryanodine receptor channel activity. Human Molecular Genetics, 2012, 21, 4497-4507.	1.4	84
30	Renpenning syndrome comes into focus. American Journal of Medical Genetics, Part A, 2005, 134A, 415-421.	0.7	78
31	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	1.8	71
32	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	0.7	68
33	Molecular Cloning and Characterization of TRPC5 (HTRP5), the Human Homologue of a Mouse Brain Receptor-Activated Capacitative Ca2+ Entry Channel. Genomics, 1999, 60, 330-340.	1.3	67
34	Novel Truncating Mutations in the Polyglutamine Tract Binding Protein 1 Gene (PQBP1) Cause Renpenning Syndrome and X-Linked Mental Retardation in Another Family with Microcephaly. American Journal of Human Genetics, 2004, 74, 777-780.	2.6	67
35	Limbâ€“body wall defect. Is there a defensible hypothesis and can it explain all the associated anomalies?. American Journal of Medical Genetics, Part A, 2011, 155, 2045-2059.	0.7	67
36	Natural history of Christianson syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2775-2783.	0.7	65

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37	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	2.6	56
38	XLMR syndrome characterized by multiple respiratory infections, hypertelorism, severe CNS deterioration and early death localizes to distal Xq28. , 1999, 85, 243-248.		55
39	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	0.7	55
40	Mutation in the 5â€² alternatively spliced region of the XNP/ATR-X gene causes Chudleyâ€“Lowry syndrome. European Journal of Human Genetics, 2005, 13, 176-183.	1.4	53
41	Affected Kindred Analysis of Human X Chromosome Exomes to Identify Novel X-Linked Intellectual Disability Genes. PLoS ONE, 2015, 10, e0116454.	1.1	49
42	ZC4H2, an XLID gene, is required for the generation of a specific subset of CNS interneurons. Human Molecular Genetics, 2015, 24, 4848-4861.	1.4	48
43	Allan-herndon-dudley syndrome: Clinical and linkage studies on a second family. American Journal of Medical Genetics Part A, 1992, 43, 491-497.	2.4	46
44	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
45	Splitting and lumping in the nosology of XLMR. American Journal of Medical Genetics Part A, 2000, 97, 174-182.	2.4	41
46	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	0.7	40
47	Abnormalities in the genes that encode Large Amino Acid Transporters increase the risk of Autism Spectrum Disorder. Molecular Genetics & Genomic Medicine, 2020, 8, e1036.	0.6	40
48	Neural tube defects and associated anomalies in South Carolina. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 554-558.	1.6	39
49	HUWE1 mutations in Juberg-Marsidi and Brooks syndromes: the results of an X-chromosome exome sequencing study. BMJ Open, 2016, 6, e009537.	0.8	39
50	Genetic syndromes among individuals with mental retardation. American Journal of Medical Genetics Part A, 2003, 123A, 29-32.	2.4	38
51	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. Nature Communications, 2020, 11, 3698.	5.8	38
52	Gastroschisis: Clinical presentation and associations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2008, 148C, 219-230.	0.7	34
53	Novel pathogenic variants in FOXP3 in fetuses with echogenic bowel and skin desquamation identified by ultrasound. , 2017, 173, 1219-1225.		31
54	Renpenning Syndrome Maps to Xp11. American Journal of Human Genetics, 1998, 62, 1092-1101.	2.6	28

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55	Holmes-Gang syndrome is allelic with XLMR-hypotonic face syndrome. American Journal of Medical Genetics Part A, 2000, 94, 383-385.	2.4	28
56	Long Term Maintenance of Neural Tube Defects Prevention in a High Prevalence State. Journal of Pediatrics, 2011, 159, 143-149.e2.	0.9	26
57	Spastic paraplegia with iron deposits in the basal ganglia: A new X-linked mental retardation syndrome. American Journal of Medical Genetics Part A, 1992, 43, 479-490.	2.4	24
58	X-linked mental retardation syndrome with short stature, small hands and feet, seizures, cleft palate, and glaucoma is linked to Xq28. , 1999, 85, 236-242.		24
59	THE IMPACT OF PRENATAL DIAGNOSIS ON NTD SURVEILLANCE. , 1996, 16, 531-535.		22
60	Hypothesis: Dysregulation of methylation of brainâ€expressed genes on the X chromosome and autism spectrum disorders. American Journal of Medical Genetics, Part A, 2008, 146A, 2213-2220.	0.7	22
61	Advances in X-linked mental retardation. Current Opinion in Pediatrics, 2005, 17, 720-724.	1.0	17
62	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)?. Molecular Genetics & Genomic Medicine, 2015, 3, 424-432.	0.6	17
63	X chromosome cDNA microarray screening identifies a functional PLP2 promoter polymorphism enriched in patients with X-linked mental retardation. Genome Research, 2007, 17, 641-648.	2.4	16
64	Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950.	0.7	16
65	Beyond osteogenesis imperfecta: Causes of fractures during infancy and childhood. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 314-327.	0.7	15
66	A Rare De Novo RAI1 Gene Mutation Affecting BDNF-Enhancer-Driven Transcription Activity Associated with Autism and Atypical Smith-Magenis Syndrome Presentation. Biology, 2018, 7, 31.	1.3	14
67	Duplication of OCRL and adjacent genes associated with autism but not Lowe syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2602-2605.	0.7	13
68	When folic acid fails: Insights from 20 years of neural tube defect surveillance in South Carolina. American Journal of Medical Genetics, Part A, 2015, 167, 2244-2250.	0.7	13
69	Ocular manifestations in the X-linked intellectual disability syndromes. Ophthalmic Genetics, 2017, 38, 401-412.	0.5	12
70	Xâ€linked intellectual disability: Phenotypic expression in carrier females. Clinical Genetics, 2020, 97, 418-425.	1.0	12
71	Common pathogenesis for sirenomelia, <scp>OEIS</scp> complex, <scp>limbâ€body</scp> wall defect, and other malformations of caudal structures. American Journal of Medical Genetics, Part A, 2021, 185, 1379-1387.	0.7	11
72	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	1.7	11

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73	Clinical geneticists in birth defects surveillance and epidemiology research programs: Past, present and future roles. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 69-75.	1.6	10
74	Clarkâ€Baraitser syndrome is associated with a nonsense alteration in the autosomal gene TRIP12. American Journal of Medical Genetics, Part A, 2020, 182, 595-596.	0.7	10
75	Neural Tube Defects and Associated Anomalies before and after Folic Acid Fortification. Journal of Pediatrics, 2020, 226, 186-194.e4.	0.9	10
76	Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. International Journal of Molecular Sciences, 2021, 22, 1111.	1.8	10
77	Prevalence of aneuploidies in South Carolina in the 1990s. Genetics in Medicine, 2002, 4, 131-135.	1.1	9
78	Shashi XLMR syndrome: Report of a second family. , 2003, 118A, 49-51.		8
79	Finding new etiologies of mental retardation and hypotonia: X marks the spot. Developmental Medicine and Child Neurology, 2008, 50, 104-111.	1.1	8
80	Seizures and X-linked intellectual disability. European Journal of Medical Genetics, 2012, 55, 307-312.	0.7	8
81	Malformations among the Xâ€linked intellectual disability syndromes. American Journal of Medical Genetics, Part A, 2013, 161, 2741-2749.	0.7	8
82	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711. European Journal of Human Genetics, 2022, 30, 420-427.	1.4	7
83	Biallelic deletions of the Waardenburg II syndrome gene, SOX10, cause a recognizable arthrogryposis syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1968-1971.	0.7	6
84	Autistic Disorder: A 20 Year Chronicle. Journal of Autism and Developmental Disorders, 2021, 51, 677-684.	1.7	5
85	Schimke XLID syndrome results from a deletion in BCAP31. American Journal of Medical Genetics, Part A, 2020, 182, 2168-2174.	0.7	4
86	Fatal hyperkeratosis syndrome in four siblings due to dolichol kinase deficiency. American Journal of Medical Genetics, Part A, 2020, 182, 1421-1425.	0.7	4
87	Arena syndrome is caused by a missense mutation in PLP1. American Journal of Medical Genetics, Part A, 2009, 149A, 1081-1081.	0.7	3
88	Hydrocephaly associated with compound heterozygous alterations in TRAPPC12. Birth Defects Research, 2020, 112, 1028-1034.	0.8	3
89	A <scp>SOX3</scp> duplication and lumbosacral spina bifida in three generations. American Journal of Medical Genetics, Part A, 2022, 188, 1572-1577.	0.7	3
90	Craniofacioskeletal syndrome: An Xâ€linked dominant disorder with early lethality in males. American Journal of Medical Genetics, Part A, 2007, 143A, 2321-2329.	0.7	2

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91	In search of the earliest images of symmelia in works of art. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 151-156.	0.7	1
92	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
93	A new test for autism spectrum disorder: Metabolic data from different cell types. Data in Brief, 2021, 39, 107598.	0.5	0