

# Roger E Stevenson

## List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

92  
papers

6,401  
citations

38  
h-index

79  
g-index

94  
ext. papers

7,279  
ext. citations

8  
avg, IF

4.83  
L-index

#	Paper	IF	Citations
92	AGTR2 mutations in X-linked mental retardation. <i>Science</i> , <b>2002</b> , 296, 2401-3	33.3	599
91	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. <i>Nature Genetics</i> , <b>2008</b> , 40, 322-8	36.3	463
90	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. <i>Nature Genetics</i> , <b>2009</b> , 41, 535-43	36.3	454
89	X-linked spastic paraplegia (SPG1), MASA syndrome and X-linked hydrocephalus result from mutations in the L1 gene. <i>Nature Genetics</i> , <b>1994</b> , 7, 402-7	36.3	377
88	Autism and maternally derived aberrations of chromosome 15q. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 76, 327-36		279
87	Evaluation of mental retardation: recommendations of a Consensus Conference: American College of Medical Genetics. <i>American Journal of Medical Genetics Part A</i> , <b>1997</b> , 72, 468-77		272
86	Allan-Herndon-Dudley syndrome and the monocarboxylate transporter 8 (MCT8) gene. <i>American Journal of Human Genetics</i> , <b>2005</b> , 77, 41-53	11	265
85	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. <i>Nature Genetics</i> , <b>2007</b> , 39, 1127-33	36.3	189
84	SLC9A6 mutations cause X-linked mental retardation, microcephaly, epilepsy, and ataxia, a phenotype mimicking Angelman syndrome. <i>American Journal of Human Genetics</i> , <b>2008</b> , 82, 1003-10	11	175
83	Fragile X and X-linked intellectual disability: four decades of discovery. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 579-90	11	166
82	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. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 345-52	11	163
81	Recurrent infections, hypotonia, and mental retardation caused by duplication of MECP2 and adjacent region in Xq28. <i>Pediatrics</i> , <b>2006</b> , 118, e1687-95	7.4	158
80	A recurrent mutation in MED12 leading to R961W causes Opitz-Kaveggia syndrome. <i>Nature Genetics</i> , <b>2007</b> , 39, 451-3	36.3	157
79	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. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 1019-27	5.6	152
78	Mutations in the DLG3 gene cause nonsyndromic X-linked mental retardation. <i>American Journal of Human Genetics</i> , <b>2004</b> , 75, 318-24	11	134
77	Mutations in the planar cell polarity genes CELSR1 and SCRIB are associated with the severe neural tube defect craniorachischisis. <i>Human Mutation</i> , <b>2012</b> , 33, 440-7	4.7	128
76	The original Lujan syndrome family has a novel missense mutation (p.N1007S) in the MED12 gene. <i>Journal of Medical Genetics</i> , <b>2007</b> , 44, 472-7	5.8	126

75	Mutations in the guanine nucleotide exchange factor gene IQSEC2 cause nonsyndromic intellectual disability. <i>Nature Genetics</i> , <b>2010</b> , 42, 486-8	36.3	118
74	Autism and macrocephaly. <i>Lancet, The</i> , <b>1997</b> , 349, 1744-5	40	117
73	A mixed epigenetic/genetic model for oligogenic inheritance of autism with a limited role for UBE3A. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 131, 1-10		112
72	X-linked spermine synthase gene (SMS) defect: the first polyamine deficiency syndrome. <i>European Journal of Human Genetics</i> , <b>2003</b> , 11, 937-44	5.3	106
71	Decreased tryptophan metabolism in patients with autism spectrum disorders. <i>Molecular Autism</i> , <b>2013</b> , 4, 16	6.5	97
70	Fine-scale survey of X chromosome copy number variants and indels underlying intellectual disability. <i>American Journal of Human Genetics</i> , <b>2010</b> , 87, 173-88	11	93
69	Mutations in ionotropic AMPA receptor 3 alter channel properties and are associated with moderate cognitive impairment in humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2007</b> , 104, 18163-8	11.5	74
68	Renpenning syndrome comes into focus. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 134, 415-21	2.5	68
67	Mutations in the BRWD3 gene cause X-linked mental retardation associated with macrocephaly. <i>American Journal of Human Genetics</i> , <b>2007</b> , 81, 367-74	11	66
66	An X-linked channelopathy with cardiomegaly due to a CLIC2 mutation enhancing ryanodine receptor channel activity. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 4497-507	5.6	64
65	Molecular cloning and characterization of TRPC5 (HTRP5), the human homologue of a mouse brain receptor-activated capacitative Ca <sup>2+</sup> entry channel. <i>Genomics</i> , <b>1999</b> , 60, 330-40	4.3	61
64	X-linked intellectual disability update 2017. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1375-1388	5.1	57
63	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. <i>American Journal of Human Genetics</i> , <b>2004</b> , 74, 777-80	11	57
62	Natural history of Christianson syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 2775-83	2.5	54
61	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , <b>2020</b> , 106, 356-370	11	51
60	Limb-body wall defect. Is there a defensible hypothesis and can it explain all the associated anomalies?. <i>American Journal of Medical Genetics, Part A</i> , <b>2011</b> , 155A, 2045-59	2.5	51
59	Mutation in the 5' alternatively spliced region of the XNP/ATR-X gene causes Chudley-Lowry syndrome. <i>European Journal of Human Genetics</i> , <b>2005</b> , 13, 176-83	5.3	45
58	Noonan syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 2323-2334	2.5	43

57	XLMR syndrome characterized by multiple respiratory infections, hypertelorism, severe CNS deterioration and early death localizes to distal Xq28. <i>American Journal of Medical Genetics Part A</i> , <b>1999</b> , 85, 243-8		43
56	Allan-Herndon-Dudley syndrome: clinical and linkage studies on a second family. <i>American Journal of Medical Genetics Part A</i> , <b>1992</b> , 43, 491-7		39
55	Affected kindred analysis of human X chromosome exomes to identify novel X-linked intellectual disability genes. <i>PLoS ONE</i> , <b>2015</b> , 10, e0116454	3.7	38
54	Splitting and lumping in the nosology of XLMR. <i>American Journal of Medical Genetics Part A</i> , <b>2000</b> , 97, 174-82		35
53	Neural tube defects and associated anomalies in South Carolina. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2004</b> , 70, 554-8		32
52	ZC4H2, an XLID gene, is required for the generation of a specific subset of CNS interneurons. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 4848-61	5.6	31
51	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1128-1136	2.5	31
50	Gastroschisis: clinical presentation and associations. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2008</b> , 148C, 219-30	3.1	31
49	Genetic syndromes among individuals with mental retardation. <i>American Journal of Medical Genetics Part A</i> , <b>2003</b> , 123A, 29-32		30
48	Gene domain-specific DNA methylation epesignatures highlight distinct molecular entities of ADNP syndrome. <i>Clinical Epigenetics</i> , <b>2019</b> , 11, 64	7.7	29
47	HUWE1 mutations in Juberg-Marsidi and Brooks syndromes: the results of an X-chromosome exome sequencing study. <i>BMJ Open</i> , <b>2016</b> , 6, e009537	3	29
46	Novel pathogenic variants in FOXP3 in fetuses with echogenic bowel and skin desquamation identified by ultrasound. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1219-1225	2.5	26
45	Renpenning syndrome maps to Xp11. <i>American Journal of Human Genetics</i> , <b>1998</b> , 62, 1092-101	11	26
44	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 150-158	2.5	25
43	Abnormalities in the genes that encode Large Amino Acid Transporters increase the risk of Autism Spectrum Disorder. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2020</b> , 8, e1036	2.3	24
42	Long term maintenance of neural tube defects prevention in a high prevalence state. <i>Journal of Pediatrics</i> , <b>2011</b> , 159, 143-149.e2	3.6	23
41	Holmes-Gang syndrome is allelic with XLMR-hypotonic face syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>2000</b> , 94, 383-5		22
40	X-linked mental retardation syndrome with short stature, small hands and feet, seizures, cleft palate, and glaucoma is linked to Xq28. <i>American Journal of Medical Genetics Part A</i> , <b>1999</b> , 85, 236-42		22

39	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. <i>American Journal of Human Genetics</i> , <b>2020</b> , 106, 234-245	11	22
38	Spastic paraplegia with iron deposits in the basal ganglia: a new X-linked mental retardation syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>1992</b> , 43, 479-90		21
37	Hypothesis: dysregulation of methylation of brain-expressed genes on the X chromosome and autism spectrum disorders. <i>American Journal of Medical Genetics, Part A</i> , <b>2008</b> , 146A, 2213-20	2.5	19
36	The impact of prenatal diagnosis on NTD surveillance. <i>Prenatal Diagnosis</i> , <b>1996</b> , 16, 531-5	3.2	18
35	X chromosome cDNA microarray screening identifies a functional PLP2 promoter polymorphism enriched in patients with X-linked mental retardation. <i>Genome Research</i> , <b>2007</b> , 17, 641-8	9.7	16
34	Advances in X-linked mental retardation. <i>Current Opinion in Pediatrics</i> , <b>2005</b> , 17, 720-4	3.2	16
33	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)?. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2015</b> , 3, 424-32	2.3	13
32	Beyond osteogenesis imperfecta: Causes of fractures during infancy and childhood. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2015</b> , 169, 314-27	3.1	13
31	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. <i>Nature Communications</i> , <b>2020</b> , 11, 3698	17.4	13
30	Ocular manifestations in the X-linked intellectual disability syndromes. <i>Ophthalmic Genetics</i> , <b>2017</b> , 38, 401-412	1.2	11
29	Duplication of OCRL and adjacent genes associated with autism but not Lowe syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 2602-5	2.5	10
28	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 1065-1074	8.1	10
27	When folic acid fails: Insights from 20 years of neural tube defect surveillance in South Carolina. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167A, 2244-50	2.5	9
26	Clinical geneticists in birth defects surveillance and epidemiology research programs: past, present and future roles. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2009</b> , 85, 69-75		8
25	Finding new etiologies of mental retardation and hypotonia: X marks the spot. <i>Developmental Medicine and Child Neurology</i> , <b>2008</b> , 50, 104-11	3.3	8
24	Shashi XLMR syndrome: report of a second family <b>2003</b> , 118A, 49-51		8
23	Seizures and X-linked intellectual disability. <i>European Journal of Medical Genetics</i> , <b>2012</b> , 55, 307-12	2.6	7
22	Prevalence of aneuploidies in South Carolina in the 1990s. <i>Genetics in Medicine</i> , <b>2002</b> , 4, 131-5	8.1	7

21	X-linked intellectual disability: Phenotypic expression in carrier females. <i>Clinical Genetics</i> , <b>2020</b> , 97, 418-425	4.25	6
20	Malformations among the X-linked intellectual disability syndromes. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 2741-9	2.5	5
19	A Rare De Novo Gene Mutation Affecting BDNF-Enhancer-Driven Transcription Activity Associated with Autism and Atypical Smith-Magenis Syndrome Presentation. <i>Biology</i> , <b>2018</b> , 7,	4.9	4
18	Biallelic deletions of the Waardenburg II syndrome gene, SOX10, cause a recognizable arthrogryposis syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1968-1971	2.5	4
17	Clark-Baraitser syndrome is associated with a nonsense alteration in the autosomal gene TRIP12. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 595-596	2.5	4
16	Rubinstein-Taybi syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 2939-2950	2.5	4
15	Arena syndrome is caused by a missense mutation in PLP1. <i>American Journal of Medical Genetics, Part A</i> , <b>2009</b> , 149A, 1081	2.5	3
14	Autistic Disorder: A 20 Year Chronicle. <i>Journal of Autism and Developmental Disorders</i> , <b>2021</b> , 51, 677-684	4.6	3
13	Fatal hyperkeratosis syndrome in four siblings due to dolichol kinase deficiency. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 1421-1425	2.5	2
12	Neural Tube Defects and Associated Anomalies before and after Folic Acid Fortification. <i>Journal of Pediatrics</i> , <b>2020</b> , 226, 186-194.e4	3.6	2
11	Craniofacioskeletal syndrome: an X-linked dominant disorder with early lethality in males. <i>American Journal of Medical Genetics, Part A</i> , <b>2007</b> , 143A, 2321-9	2.5	2
10	Schimke XLID syndrome results from a deletion in BCAP31. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 2168-2174	2.5	2
9	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711.. <i>European Journal of Human Genetics</i> , <b>2022</b> ,	5.3	1
8	Novel diagnostic DNA methylation epesignatures expand and refine the epigenetic landscapes of Mendelian disorders.. <i>Human Genetics and Genomics Advances</i> , <b>2022</b> , 3, 100075	0.8	1
7	In search of the earliest images of symmelia in works of art. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2021</b> , 187, 151-156	3.1	1
6	Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. <i>International Journal of Molecular Sciences</i> , <b>2021</b> , 22,	6.3	1
5	Hydrocephaly associated with compound heterozygous alterations in TRAPPC12. <i>Birth Defects Research</i> , <b>2020</b> , 112, 1028-1034	2.9	0
4	A SOX3 duplication and lumbosacral spina bifida in three generations.. <i>American Journal of Medical Genetics, Part A</i> , <b>2022</b> ,	2.5	0

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|---|--|-----|---|
| 3 | Deficiency of TET3 leads to a genome-wide DNA hypermethylation epesignature in human whole blood. <i>Npj Genomic Medicine</i> , <b>2021</b> , 6, 92  | 6.2 | o |
| 2 | Common pathogenesis for sirenomelia, OEIS complex, limb-body wall defect, and other malformations of caudal structures. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 1379-1387 | 2.5 | o |
| 1 | A new test for autism spectrum disorder: Metabolic data from different cell types. <i>Data in Brief</i> , <b>2021</b> , 39, 107598   | 1.2 |   |