Roger E Stevenson

List of Publications by Year in Descending Order

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	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711.. *European Journal of Human Genetics*, **2022**,	5.3	1
91	A SOX3 duplication and lumbosacral spina bifida in three generations.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
90	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1
89	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. *Npj Genomic Medicine*, **2021**, 6, 92	6.2	0
88	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	3.1	1
87	Autistic Disorder: A 20 Year Chronicle. *Journal of Autism and Developmental Disorders*, **2021**, 51, 677-684	4.6	3
86	Detection of a DNA Methylation Signature for the Intellectual Developmental Disorder, X-Linked, Syndromic, Armfield Type. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
85	Common pathogenesis for sirenomelia, OEIS complex, limb-body wall defect, and other malformations of caudal structures. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1379-1387	2.5	0
84	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. *Genetics in Medicine*, **2021**, 23, 1065-1074	8.1	10
83	A new test for autism spectrum disorder: Metabolic data from different cell types. *Data in Brief*, **2021**, 39, 107598	1.2	
82	Fatal hyperkeratosis syndrome in four siblings due to dolichol kinase deficiency. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1421-1425	2.5	2
81	Neural Tube Defects and Associated Anomalies before and after Folic Acid Fortification. *Journal of Pediatrics*, **2020**, 226, 186-194.e4	3.6	2
80	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2020**, 106, 356-370	11	51
79	Hydrocephaly associated with compound heterozygous alterations in TRAPPC12. *Birth Defects Research*, **2020**, 112, 1028-1034	2.9	0
78	Abnormalities in the genes that encode Large Amino Acid Transporters increase the risk of Autism Spectrum Disorder. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1036	2.3	24
77	X-linked intellectual disability: Phenotypic expression in carrier females. *Clinical Genetics*, **2020**, 97, 418-425	4	6
76	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. *American Journal of Human Genetics*, **2020**, 106, 234-245	11	22

75	Clark-Baraitser syndrome is associated with a nonsense alteration in the autosomal gene TRIP12. *American Journal of Medical Genetics, Part A*, **2020**, 182, 595-596	2.5	4
74	Rubinstein-Taybi syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2939-2950	2.5	4
73	Schimke XLID syndrome results from a deletion in BCAP31. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2168-2174	2.5	2
72	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. *Nature Communications*, **2020**, 11, 3698	17.4	13
71	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. *Clinical Epigenetics*, **2019**, 11, 64	7.7	29
70	Cornelia de Lange syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 150-158	2.5	25
69	Williams-Beuren syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1128-1136	2.5	31
68	X-linked intellectual disability update 2017. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1375-1388	2.5	57
67	A Rare De Novo Gene Mutation Affecting BDNF-Enhancer-Driven Transcription Activity Associated with Autism and Atypical Smith-Magenis Syndrome Presentation. *Biology*, **2018**, 7,	4.9	4
66	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	2.5	4
65	Ocular manifestations in the X-linked intellectual disability syndromes. *Ophthalmic Genetics*, **2017**, 38, 401-412	1.2	11
64	Novel pathogenic variants in FOXP3 in fetuses with echogenic bowel and skin desquamation identified by ultrasound. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1219-1225	2.5	26
63	Noonan syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2323-2334	2.5	43
62	HUWE1 mutations in Juberg-Marsidi and Brooks syndromes: the results of an X-chromosome exome sequencing study. *BMJ Open*, **2016**, 6, e009537	3	29
61	ZC4H2, an XLID gene, is required for the generation of a specific subset of CNS interneurons. *Human Molecular Genetics*, **2015**, 24, 4848-61	5.6	31
60	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, 424-32	2.3	13
59	When folic acid fails: Insights from 20 years of neural tube defect surveillance in South Carolina. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2244-50	2.5	9
58	Beyond osteogenesis imperfecta: Causes of fractures during infancy and childhood. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2015**, 169, 314-27	3.1	13

57	Affected kindred analysis of human X chromosome exomes to identify novel X-linked intellectual disability genes. *PLoS ONE*, **2015**, 10, e0116454	3.7	38
56	Decreased tryptophan metabolism in patients with autism spectrum disorders. *Molecular Autism*, **2013**, 4, 16	6.5	97
55	Malformations among the X-linked intellectual disability syndromes. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2741-9	2.5	5
54	Mutations in the planar cell polarity genes CELSR1 and SCRIB are associated with the severe neural tube defect craniorachischisis. *Human Mutation*, **2012**, 33, 440-7	4.7	128
53	Seizures and X-linked intellectual disability. *European Journal of Medical Genetics*, **2012**, 55, 307-12	2.6	7
52	Duplication of OCRL and adjacent genes associated with autism but not Lowe syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2602-5	2.5	10
51	Fragile X and X-linked intellectual disability: four decades of discovery. *American Journal of Human Genetics*, **2012**, 90, 579-90	11	166
50	An X-linked channelopathy with cardiomegaly due to a CLIC2 mutation enhancing ryanodine receptor channel activity. *Human Molecular Genetics*, **2012**, 21, 4497-507	5.6	64
49	Long term maintenance of neural tube defects prevention in a high prevalence state. *Journal of Pediatrics*, **2011**, 159, 143-149.e2	3.6	23
48	Limb-body wall defect. Is there a defensible hypothesis and can it explain all the associated anomalies?. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2045-59	2.5	51
47	Mutations in the guanine nucleotide exchange factor gene IQSEC2 cause nonsyndromic intellectual disability. *Nature Genetics*, **2010**, 42, 486-8	36.3	118
46	Fine-scale survey of X chromosome copy number variants and indels underlying intellectual disability. *American Journal of Human Genetics*, **2010**, 87, 173-88	11	93
45	Natural history of Christianson syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2775-83	2.5	54
44	Arena syndrome is caused by a missense mutation in PLP1. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1081	2.5	3
43	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		8
42	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. *Nature Genetics*, **2009**, 41, 535-43	36.3	454
41	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. *Nature Genetics*, **2008**, 40, 322-8	36.3	463
40	Finding new etiologies of mental retardation and hypotonia: X marks the spot. *Developmental Medicine and Child Neurology*, **2008**, 50, 104-11	3.3	8

39	SLC9A6 mutations cause X-linked mental retardation, microcephaly, epilepsy, and ataxia, a phenotype mimicking Angelman syndrome. *American Journal of Human Genetics*, **2008**, 82, 1003-10	11	175
38	Hypothesis: dysregulation of methylation of brain-expressed genes on the X chromosome and autism spectrum disorders. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2213-20	2.5	19
37	Gastroschisis: clinical presentation and associations. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2008**, 148C, 219-30	3.1	31
36	The original Lujan syndrome family has a novel missense mutation (p.N1007S) in the MED12 gene. *Journal of Medical Genetics*, **2007**, 44, 472-7	5.8	126
35	Craniofacioskeletal syndrome: an X-linked dominant disorder with early lethality in males. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2321-9	2.5	2
34	A recurrent mutation in MED12 leading to R961W causes Opitz-Kaveggia syndrome. *Nature Genetics*, **2007**, 39, 451-3	36.3	157
33	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. *Nature Genetics*, **2007**, 39, 1127-33	36.3	189
32	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, 18163-8	11.5	74
31	X chromosome cDNA microarray screening identifies a functional PLP2 promoter polymorphism enriched in patients with X-linked mental retardation. *Genome Research*, **2007**, 17, 641-8	9.7	16
30	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, 345-52	11	163
29	Mutations in the BRWD3 gene cause X-linked mental retardation associated with macrocephaly. *American Journal of Human Genetics*, **2007**, 81, 367-74	11	66
28	Recurrent infections, hypotonia, and mental retardation caused by duplication of MECP2 and adjacent region in Xq28. *Pediatrics*, **2006**, 118, e1687-95	7.4	158
27	Allan-Herndon-Dudley syndrome and the monocarboxylate transporter 8 (MCT8) gene. *American Journal of Human Genetics*, **2005**, 77, 41-53	11	265
26	Advances in X-linked mental retardation. *Current Opinion in Pediatrics*, **2005**, 17, 720-4	3.2	16
25	Mutation in the 5Ralternatively spliced region of the XNP/ATR-X gene causes Chudley-Lowry syndrome. *European Journal of Human Genetics*, **2005**, 13, 176-83	5.3	45
24	Renpenning syndrome comes into focus. *American Journal of Medical Genetics, Part A*, **2005**, 134, 415-21	2.5	68
23	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, 1019-27	5.6	152
22	Neural tube defects and associated anomalies in South Carolina. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2004**, 70, 554-8		32

21	A mixed epigenetic/genetic model for oligogenic inheritance of autism with a limited role for UBE3A. *American Journal of Medical Genetics Part A*, **2004**, 131, 1-10		112
20	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, 777-80	11	57
19	Mutations in the DLG3 gene cause nonsyndromic X-linked mental retardation. *American Journal of Human Genetics*, **2004**, 75, 318-24	11	134
18	Shashi XLMR syndrome: report of a second family **2003**, 118A, 49-51		8
17	Genetic syndromes among individuals with mental retardation. *American Journal of Medical Genetics Part A*, **2003**, 123A, 29-32		30
16	X-linked spermine synthase gene (SMS) defect: the first polyamine deficiency syndrome. *European Journal of Human Genetics*, **2003**, 11, 937-44	5.3	106
15	Prevalence of aneuploidies in South Carolina in the 1990s. *Genetics in Medicine*, **2002**, 4, 131-5	8.1	7
14	AGTR2 mutations in X-linked mental retardation. *Science*, **2002**, 296, 2401-3	33.3	599
13	Holmes-Gang syndrome is allelic with XLMR-hypotonic face syndrome. *American Journal of Medical Genetics Part A*, **2000**, 94, 383-5		22
12	Splitting and lumping in the nosology of XLMR. *American Journal of Medical Genetics Part A*, **2000**, 97, 174-82		35
11	X-linked mental retardation syndrome with short stature, small hands and feet, seizures, cleft palate, and glaucoma is linked to Xq28. *American Journal of Medical Genetics Part A*, **1999**, 85, 236-42		22
10	XLMR syndrome characterized by multiple respiratory infections, hypertelorism, severe CNS deterioration and early death localizes to distal Xq28. *American Journal of Medical Genetics Part A*, **1999**, 85, 243-8		43
9	Molecular cloning and characterization of TRPC5 (HTRP5), the human homologue of a mouse brain receptor-activated capacitative Ca2+ entry channel. *Genomics*, **1999**, 60, 330-40	4.3	61
8	Autism and maternally derived aberrations of chromosome 15q. *American Journal of Medical Genetics Part A*, **1998**, 76, 327-36		279
7	Renpenning syndrome maps to Xp11. *American Journal of Human Genetics*, **1998**, 62, 1092-101	11	26
6	Autism and macrocephaly. *Lancet, The*, **1997**, 349, 1744-5	40	117
5	Evaluation of mental retardation: recommendations of a Consensus Conference: American College of Medical Genetics. *American Journal of Medical Genetics Part A*, **1997**, 72, 468-77		272
4	The impact of prenatal diagnosis on NTD surveillance. *Prenatal Diagnosis*, **1996**, 16, 531-5	3.2	18

3	X-linked spastic paraplegia (SPG1), MASA syndrome and X-linked hydrocephalus result from mutations in the L1 gene. *Nature Genetics*, **1994**, 7, 402-7	36.3	377
2	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, 479-90		21
1	Allan-Herndon-Dudley syndrome: clinical and linkage studies on a second family. *American Journal of Medical Genetics Part A*, **1992**, 43, 491-7		39