Stephen P Robertson

List of Publications by Year
in descending order

95	4,719	147801 31	106344 65
papers	citations	h-index	g-index

96	96	96	7332
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	1.2	573
2	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	1.2	431
3	Localized mutations in the gene encoding the cytoskeletal protein filamin A cause diverse malformations in humans. Nature Genetics, 2003, 33, 487-491.	21.4	375
4	Identification and Successful Negotiation of a Metabolic Checkpoint in Direct Neuronal Reprogramming. Cell Stem Cell, 2016, 18, 396-409.	11.1	307
5	Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 2004, 36, 405-410.	21.4	252
6	Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. Nature Genetics, 2013, 45, 1300-1308.	21.4	247
7	Filamin A: phenotypic diversity. Current Opinion in Genetics and Development, 2005, 15, 301-307.	3.3	180
8	Germline mutations in WTX cause a sclerosing skeletal dysplasia but do not predispose to tumorigenesis. Nature Genetics, 2009, 41, 95-100.	21.4	178
9	Altered neuronal migratory trajectories in human cerebral organoids derived from individuals with neuronal heterotopia. Nature Medicine, 2019, 25, 561-568.	30.7	135
10	Gain-of-function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions. Nature Genetics, 2019, 51, 96-105.	21.4	110
11	Vascular and connective tissue anomalies associated with X-linked periventricular heterotopia due to mutations in Filamin A. European Journal of Human Genetics, 2013, 21, 494-502.	2.8	89
12	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
13	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
14	Mutations in DVL1 Cause an Osteosclerotic Form of Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 623-630.	6.2	73
15	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077.	12.8	72
16	Otopalatodigital syndrome spectrum disorders: otopalatodigital syndrome types 1 and 2, frontometaphyseal dysplasia and Melnick-Needles syndrome. European Journal of Human Genetics, 2007, 15, 3-9.	2.8	70
17	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity. American Journal of Medical Genetics, Part A, 2006, 140A, 1726-1736.	1.2	67
18	Diffuse Abnormal Layering of Small Intestinal Smooth Muscle is Present in Patients With FLNA Mutations and X-linked Intestinal Pseudo-obstruction. American Journal of Surgical Pathology, 2010, 34, 1528-1543.	3.7	64

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19	Skeletal dysplasias due to filamin A mutations result from a gain-of-function mechanism distinct from allelic neurological disordersâ€. Human Molecular Genetics, 2009, 18, 4791-4800.	2.9	63
20	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. American Journal of Human Genetics, 2016, 99, 392-406.	6.2	52
21	Terminal Osseous Dysplasia Is Caused by a Single Recurrent Mutation in the FLNA Gene. American Journal of Human Genetics, 2010, 87, 146-153.	6.2	50
22	Mutations in the netrin-1 gene cause congenital mirror movements. Journal of Clinical Investigation, 2017, 127, 3923-3936.	8.2	48
23	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. American Journal of Human Genetics, 2017, 101, 139-148.	6.2	45
24	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. Cell Reports, 2018, 25, 2729-2741.e6.	6.4	43
25	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
26	Brain dopamineâ€serotonin vesicular transport disease presenting as a severe infantile hypotonic parkinsonian disorder. Journal of Inherited Metabolic Disease, 2016, 39, 305-308.	3.6	41
27	Are Melnick-Needles syndrome and oto-palato-digital syndrome type II allelic? Observations in a four-generation kindred. , 1997, 71, 341-347.		40
28	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	3.5	40
29	<scp>ECE</scp>2 regulates neurogenesis and neuronal migration during human cortical development. EMBO Reports, 2020, 21, e48204.	4.5	40
30	Van Maldergem syndrome: further characterisation and evidence for neuronal migration abnormalities and autosomal recessive inheritance. European Journal of Human Genetics, 2012, 20, 1024-1031.	2.8	39
31	Postzygotic mutation and germline mosaicism in the otopalatodigital syndrome spectrum disorders. European Journal of Human Genetics, 2006, 14, 549-554.	2.8	38
32	Current Progress on MicroRNA-Based Gene Delivery in the Treatment of Osteoporosis and Osteoporotic Fracture. International Journal of Endocrinology, 2019, 2019, 1-17.	1.5	34
33	WTX mutations can occur both early and late in the pathogenesis of Wilms tumour. Journal of Medical Genetics, 2010, 47, 791-794.	3.2	33
34	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. Neurology, 2012, 79, 1244-1251.	1.1	31
35	The Xâ€linked filaminopathies: Synergistic insights from clinical and molecular analysis. Human Mutation, 2020, 41, 865-883.	2.5	30
36	Molecular pathology of filamin A: diverse phenotypes, many functions. Clinical Dysmorphology, 2004, 13, 123-131.	0.3	28

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37	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. Science, 2022, 376, .	12.6	25
38	Neuropsychiatric Disease in Patients With Periventricular Heterotopia. Journal of Neuropsychiatry and Clinical Neurosciences, 2013, 25, 26-31.	1.8	24
39	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	1.2	24
40	Differential regulation of twoFLNAtranscripts explains some of the phenotypic heterogeneity in the loss-of-function filaminopathies. Human Mutation, 2018, 39, 103-113.	2.5	24
41	Expanding the molecular and clinical phenotypes of FUT8â€CDG. Journal of Inherited Metabolic Disease, 2020, 43, 871-879.	3.6	24
42	Mob2 Insufficiency Disrupts Neuronal Migration in the Developing Cortex. Frontiers in Cellular Neuroscience, 2018, 12, 57.	3.7	23
43	Linkage of Otopalatodigital Syndrome Type 2 (OPD2) to Distal Xq28: Evidence for Allelism with OPD1. American Journal of Human Genetics, 2001, 69, 223-227.	6.2	22
44	Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847.	6.2	22
45	Compound Heterozygous Inheritance of Mutations in Coenzyme Q8A Results in Autosomal Recessive Cerebellar Ataxia and Coenzyme Q10 Deficiency in a Female Sib-Pair. JIMD Reports, 2017, 42, 31-36.	1.5	21
46	Extracellular LGALS3BP regulates neural progenitor position and relates to human cortical complexity. Nature Communications, 2021, 12, 6298.	12.8	21
47	Recessive Spondylocarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. American Journal of Human Genetics, 2018, 102, 1115-1125.	6.2	18
48	Biallelic variants in EFEMP1 in a man with a pronounced connective tissue phenotype. European Journal of Human Genetics, 2020, 28, 445-452.	2.8	17
49	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
50	Biallelic mutations in CYP26B1: A differential diagnosis for Pfeiffer and Antleyâ€“Bixler syndromes. American Journal of Medical Genetics, Part A, 2016, 170, 2706-2710.	1.2	15
51	Ethnic Disparity in the Incidence and Outcome of Biliary Atresia in New Zealand. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, 218-221.	1.8	15
52	Quantification of transmission risk in a male patient with a FLNB mosaic mutation causing Larsen syndrome: Implications for genetic counseling in postzygotic mosaicism cases. Human Mutation, 2017, 38, 1360-1364.	2.5	14
53	A new acro-osteolysis syndrome caused by duplications including PTHLH. Journal of Human Genetics, 2014, 59, 484-487.	2.3	13
54	The filamin-Bâ€“refilin axis â€“ spatiotemporal regulators of the actin-cytoskeleton in development and disease. Journal of Cell Science, 2018, 131, .	2.0	13

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55	Genomic medicine must reduce, not compound, health inequities: the case for hauora-enhancing genomic resources for New Zealand. New Zealand Medical Journal, 2018, 131, 81-89.	0.5	13
56	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
57	Fibulin-3 knockout mice demonstrate corneal dysfunction but maintain normal retinal integrity. Journal of Molecular Medicine, 2020, 98, 1639-1656.	3.9	11
58	Filamin A mutation associated with normal reading skills and dyslexia in a family with periventricular heterotopia. American Journal of Medical Genetics, Part A, 2012, 158A, 1897-1901.	1.2	10
59	Severe osteopathia striata with cranial sclerosis in a female case with whole WTX gene deletion. American Journal of Medical Genetics, Part A, 2013, 161, 594-599.	1.2	10
60	A Turner Syndrome Patient Carrying a Mosaic Distal X Chromosome Marker. Case Reports in Genetics, 2014, 2014, 1-5.	0.2	10
61	Association of mutations in FLNA with craniosynostosis. European Journal of Human Genetics, 2015, 23, 1684-1688.	2.8	10
62	Biallelic lossâ€ofâ€function variants in TBC1D2B cause a neurodevelopmental disorder with seizures and gingival overgrowth. Human Mutation, 2020, 41, 1645-1661.	2.5	10
63	Wilms tumor in patients with osteopathia striata with cranial sclerosis. European Journal of Human Genetics, 2021, 29, 396-401.	2.8	10
64	Int22h1/Int22h2 â€mediated Xq28 duplication syndrome: de novo duplications, prenatal diagnoses, and additional phenotypic features. Human Mutation, 2020, 41, 1238-1249.	2.5	9
65	Diverse phenotypic consequences of mutations affecting the C-terminus of FLNA. Journal of Molecular Medicine, 2015, 93, 773-782.	3.9	8
66	Frontometaphyseal dysplasia and keloid formation without FLNA mutations. American Journal of Medical Genetics, Part A, 2015, 167, 1215-1222.	1.2	8
67	Cerebral infarction in Noonan syndrome. , 1997, 71, 111-114.		7
68	Genetic investigation into an increased susceptibility to biliary atresia in an extended New Zealand MÄori family. BMC Medical Genomics, 2018, 11, 121.	1.5	7
69	Missense Pathogenic variants in KIF4A Affect Dental Morphogenesis Resulting in X-linked Taurodontism, Microdontia and Dens-Invaginatus. Frontiers in Genetics, 2019, 10, 800.	2.3	7
70	Deletion of Exon 1 in AMER1 in Osteopathia Striata with Cranial Sclerosis. Genes, 2020, 11, 1439.	2.4	7
71	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	1.3	7
72	Pathogenic variants causing ABL1 malformation syndrome cluster in a myristoyl-binding pocket and increase tyrosine kinase activity. European Journal of Human Genetics, 2021, 29, 593-603.	2.8	7

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73	An Activating Variant inCTNNB1is Associated with a Sclerosing Bone Dysplasia and Adrenocortical Neoplasia. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 688-695.	3.6	7
74	Clinical findings and a DNA methylation signature in kindreds with alterations in ZNF711. European Journal of Human Genetics, 2022, 30, 420-427.	2.8	7
75	A recurrent mutation causing Melnickâ€Needles syndrome in females confers a severe, lethal phenotype in males. American Journal of Medical Genetics, Part A, 2018, 176, 980-984.	1.2	6
76	Deletion of the last two exons of FGF10 in a family with LADD syndrome and pulmonary acinar hypoplasia. European Journal of Human Genetics, 2022, 30, 480-484.	2.8	6
77	Menkes disease: importance of diagnosis with molecular analysis in the neonatal period. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2015, 61, 407-410.	0.7	5
78	Telomere length and periodontal attachment loss: a prospective cohort study. Journal of Clinical Periodontology, 2016, 43, 121-127.	4.9	5
79	Structural and thermodynamic basis of a frontometaphyseal dysplasia mutation in filamin A. Journal of Biological Chemistry, 2017, 292, 8390-8400.	3.4	5
80	Does brittle cornea syndrome have a bone fragility phenotype?. Bone Reports, 2021, 15, 101124.	0.4	4
81	Intervertebral disc degeneration is rescued by TGFÎ²/BMP signaling modulation in an ex vivo filamin B mouse model. Bone Research, 2022, 10, 37.	11.4	4
82	Whole Exome Sequencing Reveals Compound Heterozygosity for Ethnically Distinct PEX7 Mutations Responsible for Rhizomelic Chondrodysplasia Punctata, Type 1. Case Reports in Genetics, 2015, 2015, 1-4.	0.2	3
83	Exon skipâ€inducing variants in <scp>FLNA</scp> in an attenuated form of frontometaphyseal dysplasia. American Journal of Medical Genetics, Part A, 2021, 185, 3675-3682.	1.2	3
84	Longitudinal Studies of Gene-Environment Interaction in Common Diseases-Good Value for Money?. Novartis Foundation Symposium, 2008, 293, 128-142.	1.1	3
85	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity (Am J Med Genet 140A:) Tj ETQq1 1 0.784314 rgBT /Overlock	1.2	2
86	Longitudinal studies to detect geneÃ—environment interactions in common disease â€“ Bang for your buck? A commentary on Chaufan's â€œHow much can a large population study on genes, environments, their interactions and common diseases contribute to the health of the American people?â€•(65:8,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	3.8	2
87	Recurrence of frontometaphyseal dysplasia in two sisters with a mutation in FLNA and an atypical paternal phenotype: Insights into genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2015, 167, 1161-1164.	1.2	2
88	A pilot study of exome sequencing in a diverse New Zealand cohort with undiagnosed disorders and cancer. Journal of the Royal Society of New Zealand, 2018, 48, 262-279.	1.9	2
89	The current and future state of child health and wellbeing in Aotearoa New Zealand: part 1. Journal of the Royal Society of New Zealand, 2022, 52, 313-317.	1.9	2
90	Intragenic Deletions in FLNB Are Part of the Mutational Spectrum Causing Spondylocarpotarsal Synostosis Syndrome. Genes, 2021, 12, 528.	2.4	1

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91	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	2.5	1
92	Terminal osseous dysplasia with pigmentary defects and cardiomyopathy caused by a novel FLNA variant. American Journal of Medical Genetics, Part A, 2021, 185, 3814-3820.	1.2	1
93	Filamins and Disease. , 2012, , 141-158.		1
94	Frontometaphyseal dysplasia 1 in a patient from Sri Lanka. American Journal of Medical Genetics, Part A, 2021, 185, 1317-1320.	1.2	1
95	Cantu syndrome and hypopituitarism: implications for endocrine monitoring. Endocrinology, Diabetes and Metabolism Case Reports, 2019, 2019, .	0.5	0