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	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
2	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
3	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
4	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
5	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
6	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. Science, 2022, 376, .	12.6	25
7	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
8	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
9	Wilms tumor in patients with osteopathia striata with cranial sclerosis. European Journal of Human Genetics, 2021, 29, 396-401.	2.8	10
10	Intragenic Deletions in FLNB Are Part of the Mutational Spectrum Causing Spondylocarpotarsal Synostosis Syndrome. Genes, 2021, 12, 528.	2.4	1
11	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	2.5	1
12	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
13	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
14	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
15	Does brittle cornea syndrome have a bone fragility phenotype?. Bone Reports, 2021, 15, 101124.	0.4	4
16	Frontometaphyseal dysplasia 1 in a patient from Sri Lanka. American Journal of Medical Genetics, Part A, 2021, 185, 1317-1320.	1.2	1
17	Extracellular LGALS3BP regulates neural progenitor position and relates to human cortical complexity. Nature Communications, 2021, 12, 6298.	12.8	21
18	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

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19	Fibulin-3 knockout mice demonstrate corneal dysfunction but maintain normal retinal integrity. Journal of Molecular Medicine, 2020, 98, 1639-1656.	3.9	11
20	Deletion of Exon 1 in AMER1 in Osteopathia Striata with Cranial Sclerosis. Genes, 2020, 11, 1439.	2.4	7
21	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	1.3	7
22	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
23	The Xâ€linked filaminopathies: Synergistic insights from clinical and molecular analysis. Human Mutation, 2020, 41, 865-883.	2.5	30
24	Expanding the molecular and clinical phenotypes of FUT8â€CDG. Journal of Inherited Metabolic Disease, 2020, 43, 871-879.	3.6	24
25	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
26	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
27	<scp>ECE</scp>2 regulates neurogenesis and neuronal migration during human cortical development. EMBO Reports, 2020, 21, e48204.	4.5	40
28	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
29	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	1.2	431
30	Altered neuronal migratory trajectories in human cerebral organoids derived from individuals with neuronal heterotopia. Nature Medicine, 2019, 25, 561-568.	30.7	135
31	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
32	Gain-of-function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions. Nature Genetics, 2019, 51, 96-105.	21.4	110
33	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
34	Cantu syndrome and hypopituitarism: implications for endocrine monitoring. Endocrinology, Diabetes and Metabolism Case Reports, 2019, 2019, .	0.5	0
35	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
36	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

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37	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
38	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
39	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
40	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. Cell Reports, 2018, 25, 2729-2741.e6.	6.4	43
41	Recessive Spondylocarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. American Journal of Human Genetics, 2018, 102, 1115-1125.	6.2	18
42	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
43	Mob2 Insufficiency Disrupts Neuronal Migration in the Developing Cortex. Frontiers in Cellular Neuroscience, 2018, 12, 57.	3.7	23
44	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	3.5	40
45	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
46	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	1.2	24
47	Structural and thermodynamic basis of a frontometaphyseal dysplasia mutation in filamin A. Journal of Biological Chemistry, 2017, 292, 8390-8400.	3.4	5
48	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
49	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
50	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
51	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077.	12.8	72
52	Mutations in the netrin-1 gene cause congenital mirror movements. Journal of Clinical Investigation, 2017, 127, 3923-3936.	8.2	48
53	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
54	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

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55	Telomere length and periodontal attachment loss: a prospective cohort study. Journal of Clinical Periodontology, 2016, 43, 121-127.	4.9	5
56	Identification and Successful Negotiation of a Metabolic Checkpoint in Direct Neuronal Reprogramming. Cell Stem Cell, 2016, 18, 396-409.	11.1	307
57	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
58	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
59	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
60	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
61	Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847.	6.2	22
62	Diverse phenotypic consequences of mutations affecting the C-terminus of FLNA. Journal of Molecular Medicine, 2015, 93, 773-782.	3.9	8
63	Mutations in DVL1 Cause an Osteosclerotic Form of Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 623-630.	6.2	73
64	Association of mutations in FLNA with craniosynostosis. European Journal of Human Genetics, 2015, 23, 1684-1688.	2.8	10
65	Frontometaphyseal dysplasia and keloid formation without FLNA mutations. American Journal of Medical Genetics, Part A, 2015, 167, 1215-1222.	1.2	8
66	A new acro-osteolysis syndrome caused by duplications including PTHLH. Journal of Human Genetics, 2014, 59, 484-487.	2.3	13
67	A Turner Syndrome Patient Carrying a Mosaic Distal X Chromosome Marker. Case Reports in Genetics, 2014, 2014, 1-5.	0.2	10
68	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
69	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
70	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
71	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
72	Neuropsychiatric Disease in Patients With Periventricular Heterotopia. Journal of Neuropsychiatry and Clinical Neurosciences, 2013, 25, 26-31.	1.8	24

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73	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
74	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. Neurology, 2012, 79, 1244-1251.	1.1	31
75	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
76	Filamins and Disease. , 2012, , 141-158.		1
77	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	1.2	573
78	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
79	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
80	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
81	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
82	Germline mutations in WTX cause a sclerosing skeletal dysplasia but do not predispose to tumorigenesis. Nature Genetics, 2009, 41, 95-100.	21.4	178
83	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 ETQq1 1 0.784314 rgBT /Overlock	3.8	2
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	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
86	Postzygotic mutation and germline mosaicism in the otopalatodigital syndrome spectrum disorders. European Journal of Human Genetics, 2006, 14, 549-554.	2.8	38
87	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity. American Journal of Medical Genetics, Part A, 2006, 140A, 1726-1736.	1.2	67
88	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity (Am J Med Genet 140A:) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.2	2
89	Filamin A: phenotypic diversity. Current Opinion in Genetics and Development, 2005, 15, 301-307.	3.3	180
90	Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 2004, 36, 405-410.	21.4	252

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91	Molecular pathology of filamin A: diverse phenotypes, many functions. Clinical Dysmorphology, 2004, 13, 123-131.	0.3	28
92	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
93	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
94	Cerebral infarction in Noonan syndrome. , 1997, 71, 111-114.		7
95	Are Melnick-Needles syndrome and oto-palato-digital syndrome type II allelic? Observations in a four-generation kindred. , 1997, 71, 341-347.		40