Clare Victoria Logan

List of Publications by Year
in descending order

39 papers	4,456 citations	126708 33 h-index	301761 39 g-index
40 all docs	40 docs citations	40 times ranked	7624 citing authors

#	Article	IF	Citations
1	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	2.6	352
2	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	9.4	326
3	Loss-of-function mutations in MICU1 cause a brain and muscle disorder linked to primary alterations in mitochondrial calcium signaling. Nature Genetics, 2014, 46, 188-193.	9.4	311
4	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	9.4	261
5	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	9.4	255
6	Mutations in TJP2 cause progressive cholestatic liver disease. Nature Genetics, 2014, 46, 326-328.	9.4	244
7	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
8	Mutations Causing Familial Biparental Hydatidiform Mole Implicate C6orf221 as a Possible Regulator of Genomic Imprinting in the Human Oocyte. American Journal of Human Genetics, 2011, 89, 451-458.	2.6	207
9	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 2011, 89, 713-730.	2.6	178
10	Mutations in NMNAT1 cause Leber congenital amaurosis and identify a new disease pathway for retinal degeneration. Nature Genetics, 2012, 44, 1035-1039.	9.4	177
11	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157
12	Nesprin-2 interacts with meckelin and mediates ciliogenesis via remodelling of the actin cytoskeleton. Journal of Cell Science, 2009, 122, 2716-2726.	1.2	119
13	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	9.4	118
14	Gain-of-function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions. Nature Genetics, 2019, 51, 96-105.	9.4	110
15	Identification of Mutations in SLC24A4, Encoding a Potassium-Dependent Sodium/Calcium Exchanger, as a Cause of Amelogenesis Imperfecta. American Journal of Human Genetics, 2013, 92, 307-312.	2.6	99
16	A meckelinâ€“filamin A interaction mediates ciliogenesis. Human Molecular Genetics, 2012, 21, 1272-1286.	1.4	96
17	Mutations in C4orf26, Encoding a Peptide with InÂVitro Hydroxyapatite Crystal Nucleation and Growth Activity, Cause Amelogenesis Imperfecta. American Journal of Human Genetics, 2012, 91, 565-571.	2.6	85
18	Mutations in MEGF10, a regulator of satellite cell myogenesis, cause early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). Nature Genetics, 2011, 43, 1189-1192.	9.4	84

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19	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84
20	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	9.4	81
21	Loss of the Metalloprotease ADAM9 Leads to Cone-Rod Dystrophy in Humans and Retinal Degeneration in Mice. American Journal of Human Genetics, 2009, 84, 683-691.	2.6	76
22	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	1.1	74
23	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	2.6	71
24	DNA Polymerase Epsilon Deficiency Causes IMAGe Syndrome with Variable Immunodeficiency. American Journal of Human Genetics, 2018, 103, 1038-1044.	2.6	71
25	Homozygous Mutations in PXDN Cause Congenital Cataract, Corneal Opacity, and Developmental Glaucoma. American Journal of Human Genetics, 2011, 89, 464-473.	2.6	68
26	Molecular Genetics and Pathogenic Mechanisms for the Severe Ciliopathies: Insights into Neurodevelopment and Pathogenesis of Neural Tube Defects. Molecular Neurobiology, 2011, 43, 12-26.	1.9	67
27	Next generation sequencing identifies mutations in Atonal homolog 7 (ATOH7) in families with global eye developmental defects. Human Molecular Genetics, 2012, 21, 776-783.	1.4	66
28	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	2.6	65
29	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. American Journal of Human Genetics, 2018, 103, 553-567.	2.6	58
30	Mutations in the pH-Sensing G-protein-Coupled Receptor GPR68 Cause Amelogenesis Imperfecta. American Journal of Human Genetics, 2016, 99, 984-990.	2.6	56
31	The expanding phenotype of RNU4ATAC pathogenic variants to Lowry Wood syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 465-469.	0.7	45
32	Founder mutations and genotype-phenotype correlations in Meckel-Gruber syndrome and associated ciliopathies. Cilia, 2012, 1, 18.	1.8	42
33	HACE1deficiency causes an autosomal recessive neurodevelopmental syndrome. Journal of Medical Genetics, 2015, 52, 797-803.	1.5	40
34	A homozygous STIM1 mutation impairs store-operated calcium entry and natural killer cell effector function without clinical immunodeficiency. Journal of Allergy and Clinical Immunology, 2016, 137, 955-957.e8.	1.5	38
35	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	3.8	31
36	Biallelic variants in DNA2 cause microcephalic primordial dwarfism. Human Mutation, 2019, 40, 1063-1070.	1.1	16

#	Article	IF	Citations
37	Illuminator, a desktop program for mutation detection using short-read clonal sequencing. Genomics, 2011, 98, 302-309.	1.3	5
38	Simple and Efficient Identification of Rare Recessive Pathologically Important Sequence Variants from Next Generation Exome Sequence Data. Human Mutation, 2013, 34, 945-952.	1.1	4
39	Regulation of canonical Wnt signalling by the ciliopathy protein MKS1 and the E2 ubiquitin-conjugating enzyme UBE2E1. ELife, 2022, 11, .	2.8	4