Louise S Bicknell

List of Publications by Year
in descending order

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34 papers	6,201 citations	279798 23 h-index	377865 34 g-index
34 all docs	34 docs citations	34 times ranked	10598 citing authors

#	Article	IF	Citations
1	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
2	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
3	Successful pregnancies in an adult with <scp>Meierâ€Gorlin</scp> syndrome harboring biallelic <scp>CDT1</scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 871-876.	1.2	3
4	MCM complex members MCM3 and MCM7 are associated with a phenotypic spectrum from Meier-Gorlin syndrome to lipodystrophy and adrenal insufficiency. European Journal of Human Genetics, 2021, 29, 1110-1120.	2.8	16
5	A synonymous variant in a non-canonical exon of CDC45 disrupts splicing in two affected sibs with Meier-Gorlin syndrome with craniosynostosis. European Journal of Medical Genetics, 2021, 64, 104182.	1.3	10
6	Linked-read genome sequencing identifies biallelic pathogenic variants in DONSON as a novel cause of Meier-Gorlin syndrome. Journal of Medical Genetics, 2020, 57, 195-202.	3.2	29
7	Biallelic variants in SLC35C1 as a cause of isolated short stature with intellectual disability. Journal of Human Genetics, 2020, 65, 743-750.	2.3	16
8	Expanding the phenotypic spectrum associated with DPF2: A new case report. American Journal of Medical Genetics, Part A, 2019, 179, 1637-1641.	1.2	12
9	Biallelic variants in DNA2 cause microcephalic primordial dwarfism. Human Mutation, 2019, 40, 1063-1070.	2.5	16
10	DNA Polymerase Epsilon Deficiency Causes IMAGe Syndrome with Variable Immunodeficiency. American Journal of Human Genetics, 2018, 103, 1038-1044.	6.2	71
11	Analysis of novel missense ATR mutations reveals new splicing defects underlying Seckel syndrome. Human Mutation, 2018, 39, 1847-1853.	2.5	10
12	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
13	Rare variants of the 3â€™-5â€™ DNA exonuclease TREX1 in early onset small vessel stroke. Wellcome Open Research, 2017, 2, 106.	1.8	7
14	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. American Journal of Human Genetics, 2016, 99, 125-138.	6.2	92
15	Mutations in genes encoding condensin complex proteins cause microcephaly through decatenation failure at mitosis. Genes and Development, 2016, 30, 2158-2172.	5.9	106
16	Two novel mutations in RNU4ATAC in two siblings with an atypical mild phenotype of microcephalic osteodysplastic primordial dwarfism type 1. Clinical Dysmorphology, 2016, 25, 68-72.	0.3	12
17	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. Nature Genetics, 2016, 48, 36-43.	21.4	74
18	Defective removal of ribonucleotides from DNA promotes systemic autoimmunity. Journal of Clinical Investigation, 2015, 125, 413-424.	8.2	190

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19	Mutations in the NHEJ Component XRCC4 Cause Primordial Dwarfism. American Journal of Human Genetics, 2015, 96, 412-424.	6.2	71
20	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
21	Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. Human Mutation, 2014, 35, 76-85.	2.5	74
22	Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. Nature Genetics, 2014, 46, 1283-1292.	21.4	156
23	Cerebral organoids model human brain development and microcephaly. Nature, 2013, 501, 373-379.	27.8	3,889
24	Meierâ€“Gorlin syndrome genotypeâ€“phenotype studies: 35 individuals with pre-replication complex gene mutations and 10 without molecular diagnosis. European Journal of Human Genetics, 2012, 20, 598-606.	2.8	95
25	Meierâ€“Gorlin syndrome: Growth and secondary sexual development of a microcephalic primordial dwarfism disorder. American Journal of Medical Genetics, Part A, 2012, 158A, 2733-2742.	1.2	44
26	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	21.4	201
27	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 356-359.	21.4	219
28	Mutations in ORC1, encoding the largest subunit of the origin recognition complex, cause microcephalic primordial dwarfism resembling Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 350-355.	21.4	189
29	SET Nuclear Oncogene Associates with Microcephalin/MCPH1 and Regulates Chromosome Condensation. Journal of Biological Chemistry, 2011, 286, 21393-21400.	3.4	30
30	Genetic Defects in Human Pericentrin Are Associated With Severe Insulin Resistance and Diabetes. Diabetes, 2011, 60, 925-935.	0.6	61
31	Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-Acromelic (Waardenburg Anophthalmia) Syndrome in Humans and Mice. PLoS Genetics, 2011, 7, e1002114.	3.5	81
32	A missense mutation in ALDH18A1, encoding Î”1-pyrroline-5-carboxylate synthase (P5CS), causes an autosomal recessive neurocutaneous syndrome. European Journal of Human Genetics, 2008, 16, 1176-1186.	2.8	83
33	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	3.2	102
34	Mutations in two regions ofFLNBresult in atelosteogenesis I and III. Human Mutation, 2006, 27, 705-710.	2.5	66