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	Cerebral organoids model human brain development and microcephaly. Nature, 2013, 501, 373-379.	27.8	3,889
2	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 356-359.	21.4	219
3	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	21.4	201
4	Defective removal of ribonucleotides from DNA promotes systemic autoimmunity. Journal of Clinical Investigation, 2015, 125, 413-424.	8.2	190
5	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
6	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
7	Mutations in genes encoding condensin complex proteins cause microcephaly through decatenation failure at mitosis. Genes and Development, 2016, 30, 2158-2172.	5.9	106
8	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	3.2	102
9	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
10	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
11	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
12	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
13	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
14	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
15	Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. Human Mutation, 2014, 35, 76-85.	2.5	74
16	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. Nature Genetics, 2016, 48, 36-43.	21.4	74
17	Mutations in the NHEJ Component XRCC4 Cause Primordial Dwarfism. American Journal of Human Genetics, 2015, 96, 412-424.	6.2	71
18	DNA Polymerase Epsilon Deficiency Causes IMAGe Syndrome with Variable Immunodeficiency. American Journal of Human Genetics, 2018, 103, 1038-1044.	6.2	71

#	Article	IF	Citations
19	Mutations in two regions ofFLNBresult in atelosteogenesis I and III. Human Mutation, 2006, 27, 705-710.	2.5	66
20	Genetic Defects in Human Pericentrin Are Associated With Severe Insulin Resistance and Diabetes. Diabetes, 2011, 60, 925-935.	0.6	61
21	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
22	SET Nuclear Oncogene Associates with Microcephalin/MCPH1 and Regulates Chromosome Condensation. Journal of Biological Chemistry, 2011, 286, 21393-21400.	3.4	30
23	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
24	Biallelic variants in DNA2 cause microcephalic primordial dwarfism. Human Mutation, 2019, 40, 1063-1070.	2.5	16
25	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
26	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
27	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
28	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
29	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
30	Analysis of novel missense ATR mutations reveals new splicing defects underlying Seckel syndrome. Human Mutation, 2018, 39, 1847-1853.	2.5	10
31	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
32	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
33	Rare variants of the 3â€™-5â€™ DNA exonuclease TREX1 in early onset small vessel stroke. Wellcome Open Research, 2017, 2, 106.	1.8	7
34	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