Chela James

List of Publications by Year
in descending order

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30 papers	1,246 citations	516710 16 h-index	477307 29 g-index
32 all docs	32 docs citations	32 times ranked	2393 citing authors

#	Article	IF	Citations
1	Phenotypic and Genotypic Characterisation of Inflammatory Bowel Disease Presenting Before the Age of 2 years. Journal of Crohn's and Colitis, 2017, 11, 60-69.	1.3	146
2	Hyperinsulinaemic hypoglycaemia: genetic mechanisms, diagnosis and management. Journal of Inherited Metabolic Disease, 2012, 35, 589-601.	3.6	116
3	SLC29A3 gene is mutated in pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome and interacts with the insulin signaling pathway. Human Molecular Genetics, 2009, 18, 2257-2265.	2.9	100
4	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. American Journal of Human Genetics, 2014, 95, 611-621.	6.2	89
5	Microarray analysis detects differentially expressed genes in the pharyngeal region of mice lacking Tbx1. Developmental Biology, 2005, 285, 554-569.	2.0	86
6	Advances in the diagnosis and management of hyperinsulinemic hypoglycemia. Nature Clinical Practice Endocrinology and Metabolism, 2009, 5, 101-112.	2.8	84
7	3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency and Hyperinsulinemic Hypoglycemia: Characterization of a Novel Mutation and Severe Dietary Protein Sensitivity. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 2221-2225.	3.6	72
8	A Human IPS Model Implicates Embryonic B-Myeloid Fate Restriction as Developmental Susceptibility to BÂAcute Lymphoblastic Leukemia-Associated ETV6-RUNX1. Developmental Cell, 2018, 44, 362-377.e7.	7.0	65
9	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	6.2	59
10	Hyperinsulinism in Developmental Syndromes. Endocrine Development, 2009, 14, 95-113.	1.3	56
11	Pancreatic Î²-cell KATP channels: Hypoglycaemia and hyperglycaemia. Reviews in Endocrine and Metabolic Disorders, 2010, 11, 157-163.	5.7	54
12	STAG3 truncating variant as the cause of primary ovarian insufficiency. European Journal of Human Genetics, 2016, 24, 135-138.	2.8	53
13	Genetic Analyses in Small-for-Gestational-Age Newborns. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 917-925.	3.6	38
14	Four novel cases of permanent neonatal diabetes mellitus caused by homozygous mutations in the glucokinase gene. Pediatric Diabetes, 2011, 12, 192-196.	2.9	36
15	Diabetes mellitus, exocrine pancreatic deficiency, hypertrichosis, hyperpigmentation, and chronic inflammation: confirmation of a syndrome. Pediatric Diabetes, 2009, 10, 193-197.	2.9	25
16	Chemotherapy induces canalization of cell state in childhood B-cell precursor acute lymphoblastic leukemia. Nature Cancer, 2021, 2, 835-852.	13.2	25
17	Seizures Due to a KCNQ2 Mutation: Treatment with Vitamin B6. JIMD Reports, 2015, 27, 79-84.	1.5	22
18	The onset of circulation triggers a metabolic switch required for endothelial to hematopoietic transition. Cell Reports, 2021, 37, 110103.	6.4	17

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19	Mutations in SLC25A22: hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. Journal of Inherited Metabolic Disease, 2017, 40, 385-394.	3.6	16
20	The use of whole-exome sequencing to disentangle complex phenotypes. European Journal of Human Genetics, 2016, 24, 298-301.	2.8	15
21	Generation and trapping of a mesoderm biased state of human pluripotency. Nature Communications, 2020, 11, 4989.	12.8	14
22	MicroRNA-142 Critically Regulates Group 2 Innate Lymphoid Cell Homeostasis and Function. Journal of Immunology, 2021, 206, 2725-2739.	0.8	14
23	ZSWIM7Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e254-e263.	3.6	13
24	An instructive role for Interleukin-7 receptor Î± in the development of human B-cell precursor leukemia. Nature Communications, 2022, 13, 659.	12.8	12
25	An example of the utility of genomic analysis for fast and accurate clinical diagnosis of complex rare phenotypes. Orphanet Journal of Rare Diseases, 2017, 12, 24.	2.7	10
26	A single base-pair deletion in the WFS1 gene causes Wolfram syndrome. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 389-91.	0.9	3
27	Severe Resistance to Weight Gain, Lack of Stored Triglycerides in Adipose Tissue, Hypoglycaemia, and Increased Energy Expenditure: A Novel Disorder of Energy Homeostasis. Hormone Research in Paediatrics, 2012, 77, 261-268.	1.8	3
28	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. American Journal of Human Genetics, 2015, 96, 1008-1009.	6.2	1
29	Activation of CRLF2/IL7RA Signaling in Normal Human Cord Blood Hematopoietic Progenitors Induces Philadelphia-like B-Cell Precursor Pre-Leukemia and Leukemia In Vivo. Blood, 2018, 132, 566-566.	1.4	0
30	Mitochondrial Activity Identifies a Transcriptionally and Functionally Distinct Subset of Aged HSCs with Lineage-Balanced Output. Blood, 2019, 134, 2480-2480.	1.4	0