

Chela James

List of Publications by Year in descending order

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Version: 2024-02-01

30
papers

1,246
citations

516215

16
h-index

476904

29
g-index

32
all docs

32
docs citations

32
times ranked

2393
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>ZSWIM7</i> Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e254-e263.	1.8	13
2	An instructive role for Interleukin-7 receptor $\hat{1}\pm$ in the development of human B-cell precursor leukemia. <i>Nature Communications</i> , 2022, 13, 659.	5.8	12
3	MicroRNA-142 Critically Regulates Group 2 Innate Lymphoid Cell Homeostasis and Function. <i>Journal of Immunology</i> , 2021, 206, 2725-2739.	0.4	14
4	Chemotherapy induces canalization of cell state in childhood B-cell precursor acute lymphoblastic leukemia. <i>Nature Cancer</i> , 2021, 2, 835-852.	5.7	25
5	The onset of circulation triggers a metabolic switch required for endothelial to hematopoietic transition. <i>Cell Reports</i> , 2021, 37, 110103.	2.9	17
6	Generation and trapping of a mesoderm biased state of human pluripotency. <i>Nature Communications</i> , 2020, 11, 4989.	5.8	14
7	Mitochondrial Activity Identifies a Transcriptionally and Functionally Distinct Subset of Aged HSCs with Lineage-Balanced Output. <i>Blood</i> , 2019, 134, 2480-2480.	0.6	0
8	A Human IPS Model Implicates Embryonic B-Myeloid Fate Restriction as Developmental Susceptibility to B \hat{A} Acute Lymphoblastic Leukemia-Associated ETV6-RUNX1. <i>Developmental Cell</i> , 2018, 44, 362-377.e7.	3.1	65
9	Genetic Analyses in Small-for-Gestational-Age Newborns. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 917-925.	1.8	38
10	Activation of CRLF2/IL7RA Signaling in Normal Human Cord Blood Hematopoietic Progenitors Induces Philadelphia-like B-Cell Precursor Pre-Leukemia and Leukemia In Vivo. <i>Blood</i> , 2018, 132, 566-566.	0.6	0
11	Phenotypic and Genotypic Characterisation of Inflammatory Bowel Disease Presenting Before the Age of 2 years. <i>Journal of Crohn's and Colitis</i> , 2017, 11, 60-69.	0.6	146
12	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 281-296.	2.6	59
13	An example of the utility of genomic analysis for fast and accurate clinical diagnosis of complex rare phenotypes. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 24.	1.2	10
14	Mutations in <i>SLC25A22</i> : hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 385-394.	1.7	16
15	STAG3 truncating variant as the cause of primary ovarian insufficiency. <i>European Journal of Human Genetics</i> , 2016, 24, 135-138.	1.4	53
16	The use of whole-exome sequencing to disentangle complex phenotypes. <i>European Journal of Human Genetics</i> , 2016, 24, 298-301.	1.4	15
17	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 1008-1009.	2.6	1
18	Seizures Due to a KCNQ2 Mutation: Treatment with Vitamin B6. <i>JIMD Reports</i> , 2015, 27, 79-84.	0.7	22

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19	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 611-621.	2.6	89
20	Severe Resistance to Weight Gain, Lack of Stored Triglycerides in Adipose Tissue, Hypoglycaemia, and Increased Energy Expenditure: A Novel Disorder of Energy Homeostasis. <i>Hormone Research in Paediatrics</i> , 2012, 77, 261-268.	0.8	3
21	Hyperinsulinaemic hypoglycaemia: genetic mechanisms, diagnosis and management. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 589-601.	1.7	116
22	A single base-pair deletion in the WFS1 gene causes Wolfram syndrome. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2011, 24, 389-91.	0.4	3
23	Four novel cases of permanent neonatal diabetes mellitus caused by homozygous mutations in the glucokinase gene. <i>Pediatric Diabetes</i> , 2011, 12, 192-196.	1.2	36
24	Pancreatic β -cell KATP channels: Hypoglycaemia and hyperglycaemia. <i>Reviews in Endocrine and Metabolic Disorders</i> , 2010, 11, 157-163.	2.6	54
25	SLC29A3 gene is mutated in pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome and interacts with the insulin signaling pathway. <i>Human Molecular Genetics</i> , 2009, 18, 2257-2265.	1.4	100
26	3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency and Hyperinsulinemic Hypoglycemia: Characterization of a Novel Mutation and Severe Dietary Protein Sensitivity. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 2221-2225.	1.8	72
27	Diabetes mellitus, exocrine pancreatic deficiency, hypertrichosis, hyperpigmentation, and chronic inflammation: confirmation of a syndrome. <i>Pediatric Diabetes</i> , 2009, 10, 193-197.	1.2	25
28	Hyperinsulinism in Developmental Syndromes. <i>Endocrine Development</i> , 2009, 14, 95-113.	1.3	56
29	Advances in the diagnosis and management of hyperinsulinemic hypoglycemia. <i>Nature Clinical Practice Endocrinology and Metabolism</i> , 2009, 5, 101-112.	2.9	84
30	Microarray analysis detects differentially expressed genes in the pharyngeal region of mice lacking Tbx1. <i>Developmental Biology</i> , 2005, 285, 554-569.	0.9	86