

Caroline M Joyce

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

224
citations

1307594

7
h-index

1199594

12
g-index

15
all docs

15
docs citations

15
times ranked

479
citing authors

#	ARTICLE	IF	CITATIONS
1	DNA Polymerase Epsilon Deficiency Causes IMAGE Syndrome with Variable Immunodeficiency. American Journal of Human Genetics, 2018, 103, 1038-1044.	6.2	71
2	Somatic mutations of GNA11 and GNAQ in CTNNB1-mutant aldosterone-producing adenomas presenting in puberty, pregnancy or menopause. Nature Genetics, 2021, 53, 1360-1372.	21.4	37
3	Universal Genetic Screening Uncovers a Novel Presentation of an SDHAF2 Mutation. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1392-E1396.	3.6	34
4	Establishing trimester-specific maternal thyroid function reference intervals. Annals of Clinical Biochemistry, 2014, 51, 277-283.	1.6	19
5	Mutation of a common amino acid in NKX2.5 results in dilated cardiomyopathy in two large families. BMC Medical Genetics, 2016, 17, 83.	2.1	14
6	Biallelic CYP24A1 variants presenting during pregnancy: clinical and biochemical phenotypes. Endocrine Connections, 2020, 9, 530-541.	1.9	14
7	Assessing the utility of long-read nanopore sequencing for rapid and efficient characterization of mobile element insertions. Laboratory Investigation, 2021, 101, 442-449.	3.7	9
8	Reference values for C-reactive protein and procalcitonin at term pregnancy and in the early postnatal period. Annals of Clinical Biochemistry, 2021, 58, 452-460.	1.6	9
9	The incidence of transient infantile pseudohypoadosteronism in Ireland: A prospective study. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 1257-1263.	1.5	7
10	The value of in vitro studies in a case of neonatal diabetes with a novel Kir6.2 W68G mutation. Clinical Case Reports (discontinued), 2015, 3, 884-887.	0.5	4
11	Case report of a phantom pheochromocytoma. Biochimica Medica, 2020, 30, 325-330.	2.7	4
12	Inheritance of a paternal ABCC8 variant and maternal loss of heterozygosity at 11p15 retrospectively unmasks the etiology in a case of Congenital hyperinsulinism. Clinical Case Reports (discontinued), 2020, 8, 1217-1222.	0.5	2
13	GP39...A case series of suspected congenital adrenal hyperplasia in one week in a regional endocrine unit. , 2019, , .		0
14	GP139...The incidence of transient pseudohypoadosteronism in infancy in Ireland: a prospective whole island surveillance study. , 2019, , .		0
15	Authors' reply to "Is there a role for C-reactive protein during and after labour?". Annals of Clinical Biochemistry, 2021, 58, 000456322110207.	1.6	0