

# Caroline M Joyce

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/1068032/publications.pdf>

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