Colleen M Carlston

List of Publications by Year
in descending order

13	306	1163117 8	1199594 12
papers	citations	h-index	g-index

16	16	16	977
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Loss of coordinated expression between ribosomal and mitochondrial genes revealed by comprehensive characterization of a large family with a rare Mendelian disorder. Genomics, 2021, 113, 1895-1905.	2.9	0
2	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	1.2	6
3	PQN-59 antagonizes microRNA-mediated repression during post-embryonic temporal patterning and modulates translation and stress granule formation in C. elegans. PLoS Genetics, 2021, 17, e1009599.	3.5	5
4	Diagnostic gene sequencing panels: from design to reportâ€”a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 453-461.	2.4	77
5	Baraitserâ€“Winter cerebrofrontofacial syndrome: Report of two adult siblings. American Journal of Medical Genetics, Part A, 2020, 182, 1923-1932.	1.2	2
6	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
7	Expanding the genetic and clinical spectrum of the NONOâ€associated Xâ€linked intellectual disability syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 792-796.	1.2	21
8	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
9	Three novel GJB2 (connexin 26) variants associated with autosomal dominant syndromic and nonsyndromic hearing loss. American Journal of Medical Genetics, Part A, 2018, 176, 945-950.	1.2	9
10	Extrapolation of Variant Phase in Mitochondrial Short-Chain Enoyl-CoA Hydratase (ECHS1) Deficiency. JIMD Reports, 2018, 43, 103-109.	1.5	13
11	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. Human Mutation, 2017, 38, 517-523.	2.5	49
12	The spectrum of DNMT3A variants in Tattonâ€“Brownâ€“Rahman syndrome overlaps with that in hematologic malignancies. American Journal of Medical Genetics, Part A, 2017, 173, 3022-3028.	1.2	42
13	Variable expressivity and incomplete penetrance in a large family with nonâ€classical Diamondâ€Blackfan anemia associated with ribosomal protein L11 splicing variant. American Journal of Medical Genetics, Part A, 2017, 173, 2622-2627.	1.2	14