Celia Medrano

List of Publications by Year
in descending order

8 papers	285 citations	1163117 8 h-index	1588992 8 g-index
8 all docs	8 docs citations	8 times ranked	758 citing authors

#	Article	IF	Citations
1	Clinical and molecular diagnosis of nonâ€phosphomannomutase 2 Nâ€linked congenital disorders of glycosylation in Spain. Clinical Genetics, 2019, 95, 615-626.	2.0	28
2	A Population-Based Study on Congenital Disorders of Protein N- and Combined with O-Glycosylation Experience in Clinical and Genetic Diagnosis. Journal of Pediatrics, 2017, 183, 170-177.e1.	1.8	27
3	DPAGT1-CDG: Functional analysis of disease-causing pathogenic mutations and role of endoplasmic reticulum stress. PLoS ONE, 2017, 12, e0179456.	2.5	22
4	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
5	Molecular diagnosis of glycogen storage disease and disorders with overlapping clinical symptoms by massive parallel sequencing. Genetics in Medicine, 2016, 18, 1037-1043.	2.4	32
6	Antisenseâ€mediated therapeutic pseudoexon skipping in <scp>TMEM165â€CDG</scp>. Clinical Genetics, 2015, 87, 42-48.	2.0	11
7	A novel congenital disorder of glycosylation type without central nervous system involvement caused by mutations in the phosphoglucomutase 1 gene. Journal of Inherited Metabolic Disease, 2013, 36, 535-542.	3.6	47
8	Genotypeâ€“phenotype correlations in sepiapterin reductase deficiency. A splicing defect accounts for a new phenotypic variant. Neurogenetics, 2011, 12, 183-191.	1.4	30