

Celia Medrano

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2219584/publications.pdf>

Version: 2024-02-01

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	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
2	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
3	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
4	Genotype-phenotype correlations in sepiapterin reductase deficiency. A splicing defect accounts for a new phenotypic variant. Neurogenetics, 2011, 12, 183-191.	1.4	30
5	Clinical and molecular diagnosis of non-phosphomannomutase 2 N-linked congenital disorders of glycosylation in Spain. Clinical Genetics, 2019, 95, 615-626.	2.0	28
6	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
7	DPAGT1-CDG: Functional analysis of disease-causing pathogenic mutations and role of endoplasmic reticulum stress. PLoS ONE, 2017, 12, e0179456.	2.5	22
8	Antisense-mediated therapeutic pseudoexon skipping in <sc>TMEM165</sc>-CDG. Clinical Genetics, 2015, 87, 42-48.	2.0	11