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List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9085476/publications.pdf

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22 papers	613 citations	686830 13 h-index	676716 22 g-index
22 all docs	22 docs citations	22 times ranked	876 citing authors

#	Article	IF	Citations
1	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	1.7	91
2	The challenge of CDG diagnosis. Molecular Genetics and Metabolism, 2019, 126, 1-5.	0.5	75
3	CDG Therapies: From Bench to Bedside. International Journal of Molecular Sciences, 2018, 19, 1304.	1.8	69
4	Artificial Intelligence (AI) in Rare Diseases: Is the Future Brighter?. Genes, 2019, 10, 978.	1.0	65
5	Protein misfolding diseases: Prospects of pharmacological treatment. Clinical Genetics, 2018, 93, 450-458.	1.0	44
6	The Effects of PMM2-CDG-Causing Mutations on the Folding, Activity, and Stability of the PMM2 Protein. Human Mutation, 2015, 36, 851-860.	1.1	38
7	Pharmacological Chaperoning: A Potential Treatment for PMM2-CDG. Human Mutation, 2017, 38, 160-168.	1.1	37
8	Pharmacological chaperones as a potential therapeutic option in methylmalonic aciduria cblB type. Human Molecular Genetics, 2013, 22, 3680-3689.	1.4	33
9	International consensus guidelines for phosphoglucomutase 1 deficiency (<scp>PGM1â€CDG</scp>): Diagnosis, followâ€up, and management. Journal of Inherited Metabolic Disease, 2021, 44, 148-163.	1.7	27
10	Pseudoexon exclusion by antisense therapy in 6-pyruvoyl-tetrahydropterin synthase deficiency. Human Mutation, 2011, 32, 1019-1027.	1.1	25
11	New Insights into Immunological Involvement in Congenital Disorders of Glycosylation (CDG) from a People-Centric Approach. Journal of Clinical Medicine, 2020, 9, 2092.	1.0	21
12	Patient and observer reported outcome measures to evaluate health-related quality of life in inherited metabolic diseases: a scoping review. Orphanet Journal of Rare Diseases, 2018, 13, 215.	1.2	20
13	Generation and characterization of a human iPSC line from a patient with propionic acidemia due to defects in the PCCA gene. Stem Cell Research, 2017, 23, 173-177.	0.3	14
14	Methylmalonic aciduria cblB type: characterization of two novel mutations and mitochondrial dysfunction studies. Clinical Genetics, 2015, 87, 576-581.	1.0	11
15	Artificial Intelligence in Epigenetic Studies: Shedding Light on Rare Diseases. Frontiers in Molecular Biosciences, 2021, 8, 648012.	1.6	11
16	Molecular and phenotypic characteristics of seven novel mutations causing branched-chain organic acidurias. Clinical Genetics, 2016, 90, 252-257.	1.0	10
17	New perspectives for pharmacological chaperoning treatment in methylmalonic aciduria cblB type. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 640-648.	1.8	8
18	The road to successful people-centric research in rare diseases: the web-based case study of the Immunology and Congenital Disorders of Glycosylation questionnaire (ImmunoCDGQ). Orphanet Journal of Rare Diseases, 2022, 17, 134.	1.2	4

#	Article	IF	Citations
19	Isolated and Combined Remethylation Disorders. FIRE Forum for International Research in Education, 2017, 5, 232640981668573.	0.7	3
20	Generation and characterization of two human iPSC lines from patients with methylmalonic acidemia cblB type. Stem Cell Research, 2018, 29, 143-147.	0.3	3
21	Improving the diagnosis of cobalamin and related defects by genomic analysis, plus functional and structural assessment of novel variants. Orphanet Journal of Rare Diseases, 2018, 13, 125.	1.2	3
22	A Community-Led Approach As a Guide to Overcome Challenges for Therapy Research in Congenital Disorders of Glycosylation. International Journal of Environmental Research and Public Health, 2022, 19, 6829.	1.2	1