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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	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
2	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
3	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
4	Artificial Intelligence in Epigenetic Studies: Shedding Light on Rare Diseases. Frontiers in Molecular Biosciences, 2021, 8, 648012.	1.6	11
5	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
6	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
7	Artificial Intelligence (AI) in Rare Diseases: Is the Future Brighter?. Genes, 2019, 10, 978.	1.0	65
8	The challenge of CDG diagnosis. Molecular Genetics and Metabolism, 2019, 126, 1-5.	0.5	75
9	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
10	Protein misfolding diseases: Prospects of pharmacological treatment. Clinical Genetics, 2018, 93, 450-458.	1.0	44
11	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
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	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
14	CDG Therapies: From Bench to Bedside. International Journal of Molecular Sciences, 2018, 19, 1304.	1.8	69
15	Isolated and Combined Remethylation Disorders. FIRE Forum for International Research in Education, 2017, 5, 232640981668573.	0.7	3
16	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
17	Pharmacological Chaperoning: A Potential Treatment for PMM2-CDG. Human Mutation, 2017, 38, 160-168.	1.1	37
18	Molecular and phenotypic characteristics of seven novel mutations causing branched-chain organic acidurias. Clinical Genetics, 2016, 90, 252-257.	1.0	10

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19	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
20	Methylmalonic aciduria cblB type: characterization of two novel mutations and mitochondrial dysfunction studies. Clinical Genetics, 2015, 87, 576-581.	1.0	11
21	Pharmacological chaperones as a potential therapeutic option in methylmalonic aciduria cblB type. Human Molecular Genetics, 2013, 22, 3680-3689.	1.4	33
22	Pseudoexon exclusion by antisense therapy in 6-pyruvoyl-tetrahydropterin synthase deficiency. Human Mutation, 2011, 32, 1019-1027.	1.1	25