Dulce Quelhas

List of Publications by Year
in descending order

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49 papers	1,719 citations	394421 19 h-index	276875 41 g-index
49 all docs	49 docs citations	49 times ranked	2146 citing authors

#	Article	IF	Citations
1	Glutaric Aciduria Type 2 Presenting in Adult Life With Hypoglycemia and Encephalopathic Hyperammonemia. Journal of Medical Cases, 2022, 13, 56-60.	0.7	2
2	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.	3.6	27
3	Congenital Disorders of Glycosylation in Portugalâ€”Two Decades of Experience. Journal of Pediatrics, 2021, 231, 148-156.	1.8	9
4	SLC37A4â€CDG : Second patient. JIMD Reports, 2021, 58, 122-128.	1.5	5
5	SLC35A2-CDG: Novel variant and review. Molecular Genetics and Metabolism Reports, 2021, 26, 100717.	1.1	15
6	Quantitative analysis of the natural history of prolidase deficiency: description of 17 families and systematic review of published cases. Genetics in Medicine, 2021, 23, 1604-1615.	2.4	10
7	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. Molecular Genetics and Metabolism, 2021, 133, 397-399.	1.1	3
8	Genotype-Phenotype Correlations in PMM2-CDG. Genes, 2021, 12, 1658.	2.4	6
9	Assessing the effects of PMM2 variants on protein stability. Molecular Genetics and Metabolism, 2021, 134, 344-352.	1.1	2
10	NPC1 silent variant induces skipping of exon 11 (p.V562V) and unfolded protein response was found in a specific Niemannâ€Pick type C patient. Molecular Genetics & Genomic Medicine, 2020, 8, e1451.	1.2	10
11	Consensus guideline for the diagnosis and management of mannose phosphate isomeraseâ€congenital disorder of glycosylation. Journal of Inherited Metabolic Disease, 2020, 43, 671-693.	3.6	40
12	Hyperinsulinaemic Hypoglycaemia and Polycystic Kidney Disease â€“ A Rare Case Concerning PMM2 Gene Pleiotropy. European Endocrinology, 2020, 16, 66.	1.5	7
13	Genotypeâ€phenotype correlations and BH 4 estimated responsiveness in patients with phenylketonuria from Rio de Janeiro, Southeast Brazil. Molecular Genetics & Genomic Medicine, 2019, 7, e610.	1.2	8
14	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.	3.6	91
15	Renal involvement in PMM2-CDG, a mini-review. Molecular Genetics and Metabolism, 2018, 123, 292-296.	1.1	19
16	Mutation analysis of the <scp>PAH</scp> gene in phenylketonuria patients from Rio de Janeiro, Southeast Brazil. Molecular Genetics & Genomic Medicine, 2018, 6, 575-591.	1.2	13
17	RFT1-CDG: Absence of Epilepsy and Deafness in Two Patients with Novel Pathogenic Variants. JIMD Reports, 2018, 43, 111-116.	1.5	6
18	Galactose Epimerase Deficiency: Expanding the Phenotype. JIMD Reports, 2017, 37, 19-25.	1.5	8

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19	Improving the in silico assessment of pathogenicity for compensated variants. European Journal of Human Genetics, 2017, 25, 2-7.	2.8	24
20	Mutations in the X-linked ATP6AP2 cause a glycosylation disorder with autophagic defects. Journal of Experimental Medicine, 2017, 214, 3707-3729.	8.5	62
21	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
22	Congenital disorders of glycosylation with neonatal presentation. BMJ Case Reports, 2014, 2014, bcr2013010037-bcr2013010037.	0.5	8
23	MAN1B1 Deficiency: An Unexpected CDG-II. PLoS Genetics, 2013, 9, e1003989.	3.5	63
24	Can Power Laws Help Us Understand Gene and Proteome Information?. Advances in Mathematical Physics, 2013, 2013, 1-10.	0.8	5
25	Multidimensional Scaling Applied to Histogram-Based DNA Analysis. Comparative and Functional Genomics, 2012, 2012, 1-11.	2.0	0
26	ON THE DNA OF ELEVEN MAMMALS. International Journal of Bifurcation and Chaos in Applied Sciences and Engineering, 2012, 22, 1250074.	1.7	3
27	Dietary treatment in phenylketonuria does not lead to increased risk of obesity or metabolic syndrome. Molecular Genetics and Metabolism, 2012, 107, 659-663.	1.1	69
28	Analysis and visualization of chromosome information. Gene, 2012, 491, 81-87.	2.2	5
29	Clinical, biochemical and molecular characterization of Cystinuria in a cohort of 12 patients. Clinical Genetics, 2012, 81, 47-55.	2.0	35
30	Wavelet analysis of human DNA. Genomics, 2011, 98, 155-163.	2.9	35
31	Shannon, RÃ©nyie and Tsallis entropy analysis of DNA using phase plane. Nonlinear Analysis: Real World Applications, 2011, 12, 3135-3144.	1.7	29
32	Entropy analysis of the DNA code dynamics in human chromosomes. Computers and Mathematics With Applications, 2011, 62, 1612-1617.	2.7	23
33	Fractional dynamics in DNA. Communications in Nonlinear Science and Numerical Simulation, 2011, 16, 2963-2969.	3.3	58
34	Relevance of Expanded Neonatal Screening of Medium-Chain Acyl Co-A Dehydrogenase Deficiency: Outcome of a Decade in Galicia (Spain). JIMD Reports, 2011, 1, 131-136.	1.5	2
35	The Molecular Landscape of Phosphomannose Mutase Deficiency in Iberian Peninsula: Identification of 15 Population-Specific Mutations. JIMD Reports, 2011, 1, 117-123.	1.5	16
36	Quantitative analysis of five sterols in amniotic fluid by GCâ€“MS: Application to the diagnosis of cholesterol biosynthesis defects. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2010, 878, 2130-2136.	2.3	21

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37	De Barsy syndrome and ATP6V0A2-CDG. European Journal of Human Genetics, 2010, 18, 526-526.	2.8	16
38	Golgi function and dysfunction in the first COG4-deficient CDG type II patient. Human Molecular Genetics, 2009, 18, 3244-3256.	2.9	129
39	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	21.4	330
40	Screening Using Serum Percentage of Carbohydrate-Deficient Transferrin for Congenital Disorders of Glycosylation in Children with Suspected Metabolic Disease. Clinical Chemistry, 2008, 54, 93-100.	3.2	33
41	Congenital Disorder of Glycosylation Type Ia: Searching for the Origin of Common Mutations inPMM2. Annals of Human Genetics, 2007, 71, 348-353.	0.8	13
42	Prenatal diagnosis for CDG Ia based on post-mortem molecular study of Guthrie card. Molecular Genetics and Metabolism, 2006, 87, 379.	1.1	4
43	Conserved oligomeric Golgi complex subunit 1 deficiency reveals a previously uncharacterized congenital disorder of glycosylation type II. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3764-3769.	7.1	175
44	Strenuous exercise aggravates MDMA-induced skeletal muscle damage in mice. Toxicology, 2005, 206, 349-358.	4.2	18
45	Detailed glycan analysis of serum glycoproteins of patients with congenital disorders of glycosylation indicates the specific defective glycan processing step and provides an insight into pathogenesis. Glycobiology, 2003, 13, 601-622.	2.5	138
46	X-linked adrenoleukodystrophy in patients with idiopathic addison disease. European Journal of Pediatrics, 1994, 153, 594-597.	2.7	30
47	X-linked adrenoleukodystrophy in patients with idiopathic Addison disease. European Journal of Pediatrics, 1994, 153, 594-597.	2.7	1
48	Characterization of X-linked adrenoleukodystrophy in different biological specimens from ten Portuguese families. Journal of Inherited Metabolic Disease, 1993, 16, 55-62.	3.6	3
49	X-linked adrenoleukodystrophy and haemophilia A in the same kindred. Journal of Inherited Metabolic Disease, 1993, 16, 595-598.	3.6	2