

Dulce Quelhas

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2389582/publications.pdf>

Version: 2024-02-01

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	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H ⁺ -ATPase subunit ATP6VOA2. <i>Nature Genetics</i> , 2008, 40, 32-34.	21.4	330
2	Conserved oligomeric Golgi complex subunit 1 deficiency reveals a previously uncharacterized congenital disorder of glycosylation type II. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 3764-3769.	7.1	175
3	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. <i>Glycobiology</i> , 2003, 13, 601-622.	2.5	138
4	Golgi function and dysfunction in the first COG4-deficient CDG type II patient. <i>Human Molecular Genetics</i> , 2009, 18, 3244-3256.	2.9	129
5	International clinical guidelines for the management of phosphomannomutase 2â€œcongenital disorders of glycosylation: Diagnosis, treatment and follow up. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 5-28.	3.6	91
6	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. <i>American Journal of Human Genetics</i> , 2016, 98, 310-321.	6.2	88
7	Dietary treatment in phenylketonuria does not lead to increased risk of obesity or metabolic syndrome. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 659-663.	1.1	69
8	MAN1B1 Deficiency: An Unexpected CDG-II. <i>PLoS Genetics</i> , 2013, 9, e1003989.	3.5	63
9	Mutations in the X-linked <i>ATP6AP2</i> cause a glycosylation disorder with autophagic defects. <i>Journal of Experimental Medicine</i> , 2017, 214, 3707-3729.	8.5	62
10	Fractional dynamics in DNA. <i>Communications in Nonlinear Science and Numerical Simulation</i> , 2011, 16, 2963-2969.	3.3	58
11	Consensus guideline for the diagnosis and management of mannose phosphate isomeraseâ€œcongenital disorder of glycosylation. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 671-693.	3.6	40
12	Wavelet analysis of human DNA. <i>Genomics</i> , 2011, 98, 155-163.	2.9	35
13	Clinical, biochemical and molecular characterization of Cystinuria in a cohort of 12 patients. <i>Clinical Genetics</i> , 2012, 81, 47-55.	2.0	35
14	Screening Using Serum Percentage of Carbohydrate-Deficient Transferrin for Congenital Disorders of Glycosylation in Children with Suspected Metabolic Disease. <i>Clinical Chemistry</i> , 2008, 54, 93-100.	3.2	33
15	X-linked adrenoleukodystrophy in patients with idiopathic addison disease. <i>European Journal of Pediatrics</i> , 1994, 153, 594-597.	2.7	30
16	Shannon, RÃ©nyie and Tsallis entropy analysis of DNA using phase plane. <i>Nonlinear Analysis: Real World Applications</i> , 2011, 12, 3135-3144.	1.7	29
17	International consensus guidelines for phosphoglucomutase 1 deficiency (<i>PGM1</i> â€œCDG): Diagnosis, followâ€œup, and management. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 148-163.	3.6	27
18	Improving the in silico assessment of pathogenicity for compensated variants. <i>European Journal of Human Genetics</i> , 2017, 25, 2-7.	2.8	24

#	ARTICLE	IF	CITATIONS
19	Entropy analysis of the DNA code dynamics in human chromosomes. <i>Computers and Mathematics With Applications</i> , 2011, 62, 1612-1617.	2.7	23
20	Quantitative analysis of five sterols in amniotic fluid by GC-MS: Application to the diagnosis of cholesterol biosynthesis defects. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2010, 878, 2130-2136.	2.3	21
21	Renal involvement in PMM2-CDG, a mini-review. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 292-296.	1.1	19
22	Strenuous exercise aggravates MDMA-induced skeletal muscle damage in mice. <i>Toxicology</i> , 2005, 206, 349-358.	4.2	18
23	De Barsy syndrome and ATP6V0A2-CDG. <i>European Journal of Human Genetics</i> , 2010, 18, 526-526.	2.8	16
24	The Molecular Landscape of Phosphomannose Mutase Deficiency in Iberian Peninsula: Identification of 15 Population-Specific Mutations. <i>JIMD Reports</i> , 2011, 1, 117-123.	1.5	16
25	SLC35A2-CDG: Novel variant and review. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100717.	1.1	15
26	Congenital Disorder of Glycosylation Type Ia: Searching for the Origin of Common Mutations in PMM2. <i>Annals of Human Genetics</i> , 2007, 71, 348-353.	0.8	13
27	Mutation analysis of the <i>PAH</i> gene in phenylketonuria patients from Rio de Janeiro, Southeast Brazil. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 575-591.	1.2	13
28	<i>NPC1</i> silent variant induces skipping of exon 11 (p.V562V) and unfolded protein response was found in a specific Niemann-Pick type C patient. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1451.	1.2	10
29	Quantitative analysis of the natural history of prolidase deficiency: description of 17 families and systematic review of published cases. <i>Genetics in Medicine</i> , 2021, 23, 1604-1615.	2.4	10
30	Congenital Disorders of Glycosylation in Portugal—Two Decades of Experience. <i>Journal of Pediatrics</i> , 2021, 231, 148-156.	1.8	9
31	Congenital disorders of glycosylation with neonatal presentation. <i>BMJ Case Reports</i> , 2014, 2014, bcr2013010037-bcr2013010037.	0.5	8
32	Galactose Epimerase Deficiency: Expanding the Phenotype. <i>JIMD Reports</i> , 2017, 37, 19-25.	1.5	8
33	Genotype-phenotype correlations and BH 4 estimated responsiveness in patients with phenylketonuria from Rio de Janeiro, Southeast Brazil. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e610.	1.2	8
34	Hyperinsulinaemic Hypoglycaemia and Polycystic Kidney Disease – A Rare Case Concerning <i>PMM2</i> Gene Pleiotropy. <i>European Endocrinology</i> , 2020, 16, 66.	1.5	7
35	RFT1-CDG: Absence of Epilepsy and Deafness in Two Patients with Novel Pathogenic Variants. <i>JIMD Reports</i> , 2018, 43, 111-116.	1.5	6
36	Genotype-Phenotype Correlations in PMM2-CDG. <i>Genes</i> , 2021, 12, 1658.	2.4	6

#	ARTICLE	IF	CITATIONS
37	Analysis and visualization of chromosome information. <i>Gene</i> , 2012, 491, 81-87.	2.2	5
38	Can Power Laws Help Us Understand Gene and Proteome Information?. <i>Advances in Mathematical Physics</i> , 2013, 2013, 1-10.	0.8	5
39	SLC37A4â€CDG : Second patient. <i>JIMD Reports</i> , 2021, 58, 122-128.	1.5	5
40	Prenatal diagnosis for CDG Ia based on post-mortem molecular study of Guthrie card. <i>Molecular Genetics and Metabolism</i> , 2006, 87, 379.	1.1	4
41	Characterization of X-linked adrenoleukodystrophy in different biological specimens from ten Portuguese families. <i>Journal of Inherited Metabolic Disease</i> , 1993, 16, 55-62.	3.6	3
42	ON THE DNA OF ELEVEN MAMMALS. <i>International Journal of Bifurcation and Chaos in Applied Sciences and Engineering</i> , 2012, 22, 1250074.	1.7	3
43	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. <i>Molecular Genetics and Metabolism</i> , 2021, 133, 397-399.	1.1	3
44	X-linked adrenoleukodystrophy and haemophilia A in the same kindred. <i>Journal of Inherited Metabolic Disease</i> , 1993, 16, 595-598.	3.6	2
45	Relevance of Expanded Neonatal Screening of Medium-Chain Acyl Co-A Dehydrogenase Deficiency: Outcome of a Decade in Galicia (Spain). <i>JIMD Reports</i> , 2011, 1, 131-136.	1.5	2
46	Assessing the effects of PMM2 variants on protein stability. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 344-352.	1.1	2
47	Glutaric Aciduria Type 2 Presenting in Adult Life With Hypoglycemia and Encephalopathic Hyperammonemia. <i>Journal of Medical Cases</i> , 2022, 13, 56-60.	0.7	2
48	X-linked adrenoleukodystrophy in patients with idiopathic Addison disease. <i>European Journal of Pediatrics</i> , 1994, 153, 594-597.	2.7	1
49	Multidimensional Scaling Applied to Histogram-Based DNA Analysis. <i>Comparative and Functional Genomics</i> , 2012, 2012, 1-11.	2.0	0