Olga M Amaral

List of Publications by Year
in descending order

34	830	687363 13	477307 29
papers	citations	h-index	g-index

38	38	38	1184
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence of lysosomal storage diseases in Portugal. European Journal of Human Genetics, 2004, 12, 87-92.	2.8	277
2	Niemann-Pick type C disease: NPC1 mutations associated with severe and mild cellular cholesterol trafficking alterations. Human Genetics, 2001, 109, 24-32.	3.8	90
3	Gaucher Disease: The Origins of the Ashkenazi Jewish N370S and 84GG Acid Î²-Glucosidase Mutations. American Journal of Human Genetics, 2000, 66, 1821-1832.	6.2	62
4	T Cell Numbers Relate to Bone Involvement in Gaucher Disease. Blood Cells, Molecules, and Diseases, 1999, 25, 130-138.	1.4	47
5	Type 1 Gaucher Disease: Molecular, Biochemical, and Clinical Characterization of Patients from Northern Portugal. Biochemical Medicine and Metabolic Biology, 1993, 49, 97-107.	0.7	31
6	Type 1 Gaucher disease: Identification of N396T and prevalence of glucocerebrosidase mutations in the Portuguese. Human Mutation, 1996, 8, 280-281.	2.5	29
7	Allelic frequency determination of the 24-bp chitotriosidase duplication in the Portuguese population by real-time PCR. Blood Cells, Molecules, and Diseases, 2004, 33, 362-364.	1.4	29
8	Mutations c.459+1G>A and p.P426L in the ARSA gene: Prevalence in metachromatic leukodystrophy patients from European countries. Molecular Genetics and Metabolism, 2005, 86, 353-359.	1.1	27
9	Adult-onset neuronopathic form of Gaucher's disease: a case report. Parkinsonism and Related Disorders, 2003, 9, 261-264.	2.2	26
10	Gaucher disease: expression and characterization of mild and severe acid Î²-glucosidase mutations in Portuguese type 1 patients. European Journal of Human Genetics, 2000, 8, 95-102.	2.8	25
11	Molecular characterisation of type 1 Gaucher disease families and patients: intrafamilial heterogeneity at the clinical level.. Journal of Medical Genetics, 1994, 31, 401-404.	3.2	23
12	Advances in Sphingolipidoses: CRISPR-Cas9 Editing as an Option for Modelling and Therapy. International Journal of Molecular Sciences, 2019, 20, 5897.	4.1	15
13	Molecular analysis of mucopolysaccharidosis type IIIB in Portugal: evidence of a single origin for a common mutation (R234C) in the Iberian Peninsula. Clinical Genetics, 2008, 73, 251-256.	2.0	14
14	Metachromatic leucodystrophy in Portugal-finding of four new molecular lesions: C300F, P425T, g.1190-1191insC, and g.2408delC;. Human Mutation, 1999, 13, 337-338.	2.5	13
15	Correction of a Splicing Mutation Affecting an Unverricht-Lundborg Disease Patient by Antisense Therapy. Genes, 2018, 9, 455.	2.4	13
16	In Silico Analysis of Missense Mutations as a First Step in Functional Studies: Examples from Two Sphingolipidoses. International Journal of Molecular Sciences, 2018, 19, 3409.	4.1	12
17	Homozygosity for two mild glucocerebrosidase mutations of probable Iberian origin. Clinical Genetics, 1999, 56, 100-102.	2.0	11
18	Unverrichtâ€“Lundborg disease: Homozygosity for a new splicing mutation in the cystatin B gene. Epilepsy Research, 2012, 99, 187-190.	1.6	11

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19	The N370S mutation in the glucocerebrosidase gene of Portuguese type 1 Gaucher patients: Linkage to the PvuII polymorphism. Journal of Inherited Metabolic Disease, 1994, 17, 85-88.	3.6	10
20	Gaucher disease in Tunisia: High frequency of the most common mutations. Blood Cells, Molecules, and Diseases, 2009, 43, 161-162.	1.4	9
21	Distinct Haplotype in Non-Ashkenazi Gaucher Patients with N370S Mutation. Blood Cells, Molecules, and Diseases, 1997, 23, 415-416.	1.4	8
22	Characterization of a rare Unverrichtâ€“Lundborg disease mutation. Molecular Genetics and Metabolism Reports, 2015, 4, 68-71.	1.1	8
23	Segmental and total uniparental isodisomy (UPiD) as a disease mechanism in autosomal recessive lysosomal disorders: evidence from SNP arrays. European Journal of Human Genetics, 2019, 27, 919-927.	2.8	8
24	Rapid and Cost-Effective Method for the Detection of the c.533G>A Mutation in theHEXAGene. Genetic Testing and Molecular Biomarkers, 2011, 15, 123-126.	0.7	6
25	Identification of Gaucher Disease Carriers: Glucocerebrosidase Antigen and DNA Analysis. Biochemical Medicine and Metabolic Biology, 1993, 50, 190-196.	0.7	4
26	ARSA-PD associated alleles in the Portuguese population: frequency determination and haplotype analysis. Molecular Genetics and Metabolism, 2003, 79, 305-307.	1.1	4
27	Mutation Frequency of Three Neurodegenerative Lysosomal Storage Diseases: From Screening to Treatment?. Archives of Medical Research, 2017, 48, 263-269.	3.3	4
28	Induced pluripotent stem cell line (INSAi001-A) from a Gaucher disease type 3 patient compound heterozygote for mutations in the GBA1 gene. Stem Cell Research, 2019, 41, 101595.	0.7	4
29	CHIT1 genetic defects in the Portuguese population. Blood Cells, Molecules, and Diseases, 2013, 50, 50-52.	1.4	3
30	Induced pluripotent stem cell line (INSAi002-A) from a Fabry Disease patient hemizygote for the rare p.W287X mutation. Stem Cell Research, 2020, 45, 101794.	0.7	3
31	Efficient IDUA Gene Mutation Detection with Combined Use of dHPLC and Dried Blood Samples. , 2013, 2013, 1-4.		1
32	Lessons from CiÃªncia Viva: how teaching human genetics to XXIst century students must go beyond the classroom. European Journal of Human Genetics, 2020, 28, 533-534.	2.8	0
33	CRISPR/Cas in iPSCs from sphingolipidoses patients. Molecular Genetics and Metabolism, 2019, 126, S101-S102.	1.1	0
34	Applications of iPSCs in Gaucher Disease and other rare sphingolipidoses. , 2022, , 197-224.		0