

# Olga M Amaral

## List of Publications by Year in descending order

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Version: 2024-02-01

34  
papers

830  
citations

687363

13  
h-index

477307

29  
g-index

38  
all docs

38  
docs citations

38  
times ranked

1184  
citing authors

#	ARTICLE	IF	CITATIONS
1	Applications of iPSCs in Gaucher Disease and other rare sphingolipidoses. , 2022, , 197-224.		0
2	Lessons from Ciência Viva: how teaching human genetics to XX1st century students must go beyond the classroom. European Journal of Human Genetics, 2020, 28, 533-534.	2.8	0
3	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
4	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
5	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
6	Advances in Sphingolipidoses: CRISPR-Cas9 Editing as an Option for Modelling and Therapy. International Journal of Molecular Sciences, 2019, 20, 5897.	4.1	15
7	CRISPR/Cas in iPSCs from sphingolipidoses patients. Molecular Genetics and Metabolism, 2019, 126, S101-S102.	1.1	0
8	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
9	Correction of a Splicing Mutation Affecting an Unverricht-Lundborg Disease Patient by Antisense Therapy. Genes, 2018, 9, 455.	2.4	13
10	Mutation Frequency of Three Neurodegenerative Lysosomal Storage Diseases: From Screening to Treatment?. Archives of Medical Research, 2017, 48, 263-269.	3.3	4
11	Characterization of a rare Unverricht-Lundborg disease mutation. Molecular Genetics and Metabolism Reports, 2015, 4, 68-71.	1.1	8
12	CHIT1 genetic defects in the Portuguese population. Blood Cells, Molecules, and Diseases, 2013, 50, 50-52.	1.4	3
13	Efficient IDUA Gene Mutation Detection with Combined Use of dHPLC and Dried Blood Samples. , 2013, 2013, 1-4.		1
14	Unverricht-Lundborg disease: Homozygosity for a new splicing mutation in the cystatin B gene. Epilepsy Research, 2012, 99, 187-190.	1.6	11
15	Rapid and Cost-Effective Method for the Detection of the c.533G>A Mutation in the HEXA Gene. Genetic Testing and Molecular Biomarkers, 2011, 15, 123-126.	0.7	6
16	Gaucher disease in Tunisia: High frequency of the most common mutations. Blood Cells, Molecules, and Diseases, 2009, 43, 161-162.	1.4	9
17	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
18	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

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19	Prevalence of lysosomal storage diseases in Portugal. <i>European Journal of Human Genetics</i> , 2004, 12, 87-92.	2.8	277
20	Allelic frequency determination of the 24-bp chitotriosidase duplication in the Portuguese population by real-time PCR. <i>Blood Cells, Molecules, and Diseases</i> , 2004, 33, 362-364.	1.4	29
21	ARSA-PD associated alleles in the Portuguese population: frequency determination and haplotype analysis. <i>Molecular Genetics and Metabolism</i> , 2003, 79, 305-307.	1.1	4
22	Adult-onset neuronopathic form of Gaucher's disease: a case report. <i>Parkinsonism and Related Disorders</i> , 2003, 9, 261-264.	2.2	26
23	Niemann-Pick type C disease: NPC1 mutations associated with severe and mild cellular cholesterol trafficking alterations. <i>Human Genetics</i> , 2001, 109, 24-32.	3.8	90
24	Gaucher disease: expression and characterization of mild and severe acid $\beta$ -glucosidase mutations in Portuguese type 1 patients. <i>European Journal of Human Genetics</i> , 2000, 8, 95-102.	2.8	25
25	Gaucher Disease: The Origins of the Ashkenazi Jewish N370S and 84GG Acid $\beta$ -Glucosidase Mutations. <i>American Journal of Human Genetics</i> , 2000, 66, 1821-1832.	6.2	62
26	Homozygosity for two mild glucocerebrosidase mutations of probable Iberian origin. <i>Clinical Genetics</i> , 1999, 56, 100-102.	2.0	11
27	Metachromatic leucodystrophy in Portugal-finding of four new molecular lesions: C300F, P425T, g.1190-1191insC, and g.2408delC;. <i>Human Mutation</i> , 1999, 13, 337-338.	2.5	13
28	T Cell Numbers Relate to Bone Involvement in Gaucher Disease. <i>Blood Cells, Molecules, and Diseases</i> , 1999, 25, 130-138.	1.4	47
29	Distinct Haplotype in Non-Ashkenazi Gaucher Patients with N370S Mutation. <i>Blood Cells, Molecules, and Diseases</i> , 1997, 23, 415-416.	1.4	8
30	Type 1 Gaucher disease: Identification of N396T and prevalence of glucocerebrosidase mutations in the Portuguese. <i>Human Mutation</i> , 1996, 8, 280-281.	2.5	29
31	Molecular characterisation of type 1 Gaucher disease families and patients: intrafamilial heterogeneity at the clinical level.. <i>Journal of Medical Genetics</i> , 1994, 31, 401-404.	3.2	23
32	The N370S mutation in the glucocerebrosidase gene of Portuguese type 1 Gaucher patients: Linkage to the PvuII polymorphism. <i>Journal of Inherited Metabolic Disease</i> , 1994, 17, 85-88.	3.6	10
33	Type 1 Gaucher Disease: Molecular, Biochemical, and Clinical Characterization of Patients from Northern Portugal. <i>Biochemical Medicine and Metabolic Biology</i> , 1993, 49, 97-107.	0.7	31
34	Identification of Gaucher Disease Carriers: Glucocerebrosidase Antigen and DNA Analysis. <i>Biochemical Medicine and Metabolic Biology</i> , 1993, 50, 190-196.	0.7	4