RosÃ¡rio Santos

List of Publications by Year
in descending order

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74 papers	1,821 citations	331670 21 h-index	289244 40 g-index
75 all docs	75 docs citations	75 times ranked	3034 citing authors

#	Article	IF	Citations
1	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	2.5	507
2	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	2.6	125
3	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. Human Mutation, 2013, 34, 1449-1457.	2.5	94
4	LAMA2gene mutation update: Toward a more comprehensive picture of the laminin-Î±2 variome and its related phenotypes. Human Mutation, 2018, 39, 1314-1337.	2.5	71
5	LAMA2 gene analysis in a cohort of 26 congenital muscular dystrophy patients. Clinical Genetics, 2008, 74, 502-512.	2.0	61
6	Mutation screening of the thyroid peroxidase gene in a cohort of 55 Portuguese patients with congenital hypothyroidism. European Journal of Endocrinology, 2005, 152, 193-198.	3.7	56
7	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. European Journal of Human Genetics, 2002, 10, 197-203.	2.8	45
8	Dominant and recessive RYR1 mutations in adults with core lesions and mild muscle symptoms. Muscle and Nerve, 2011, 44, 102-108.	2.2	39
9	Mutation analysis in patients with total sperm immotility. Journal of Assisted Reproduction and Genetics, 2015, 32, 893-902.	2.5	36
10	New variants, challenges and pitfalls in DMD genotyping: implications in diagnosis, prognosis and therapy. Journal of Human Genetics, 2014, 59, 454-464.	2.3	35
11	EMQN best practice guidelines for genetic testing in dystrophinopathies. European Journal of Human Genetics, 2020, 28, 1141-1159.	2.8	35
12	New splicing mutation in the choline kinase beta (CHKB) gene causing a muscular dystrophy detected by whole-exome sequencing. Journal of Human Genetics, 2015, 60, 305-312.	2.3	33
13	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
14	Atypical phenotype in two patients with LAMA2 mutations. Neuromuscular Disorders, 2014, 24, 419-424.	0.6	30
15	Expanding the MTM1 mutational spectrum: novel variants including the first multi-exonic duplication and development of a locus-specific database. European Journal of Human Genetics, 2013, 21, 540-549.	2.8	29
16	Analysis of the UDP-glucuronosyltransferase gene in Portuguese patients with a clinical diagnosis of Gilbert and Crigler–Najjar syndromes. Blood Cells, Molecules, and Diseases, 2006, 36, 91-97.	1.4	28
17	The Polymorphism c.-3279T>G in the Phenobarbital-Responsive Enhancer Module of the Bilirubin UDP-Glucuronosyltransferase Gene Is Associated with Gilbert Syndrome. Clinical Chemistry, 2005, 51, 2204-2206.	3.2	26
18	Clinical and Genetic Analysis of Children with Kartagener Syndrome. Cells, 2019, 8, 900.	4.1	26

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19	Exonization of an Intronic LINE-1 Element Causing Becker Muscular Dystrophy as a Novel Mutational Mechanism in Dystrophin Gene. Genes, 2017, 8, 253.	2.4	25
20	The new neuromuscular disease related with defects in the <scp>ASC</scp>â€1 complex: report of a second case confirms <scp>ASCC1</scp> involvement. Clinical Genetics, 2017, 92, 434-439.	2.0	23
21	Characterization of CCDC103 expression profiles: further insights in primary ciliary dyskinesia and in human reproduction. Journal of Assisted Reproduction and Genetics, 2019, 36, 1683-1700.	2.5	23
22	Analysis of Free and Total Glycerol in Biodiesel Using an Electrochemical Assay Based on a Two-Enzyme Oxygen-Electrode System. Electroanalysis, 2010, 22, 995-999.	2.9	21
23	Oxidized lowâ€density lipoprotein and lipoprotein(a) levels in chronic kidney disease patients under hemodialysis: Influence of adiponectin and of a polymorphism in the apolipoprotein(a) gene. Hemodialysis International, 2012, 16, 481-490.	0.9	21
24	Unraveling the pathogenesis of ARX polyalanine tract variants using a clinical and molecular interfacing approach. Molecular Genetics & Genomic Medicine, 2015, 3, 203-214.	1.2	21
25	Impact of UGT1A1 gene variants on total bilirubin levels in Gilbert syndrome patients and in healthy subjects. Blood Cells, Molecules, and Diseases, 2012, 48, 166-172.	1.4	20
26	Development of NIPBL Locus-Specific Database Using LOVD: From Novel Mutations to Further Genotype-Phenotype Correlations in Cornelia de Lange Syndrome. Human Mutation, 2010, 31, 1216-1222.	2.5	19
27	Adipokine Gene Single-Nucleotide Polymorphisms in Portuguese Obese Adolescents: Associations with Plasma Concentrations of Adiponectin, Resistin, IL-6, IL-1Î², and TNF-Î±. Childhood Obesity, 2016, 12, 300-313.	1.5	18
28	Contraction of fully expanded FMR1 alleles to the normal range: predisposing haplotype or rare events?. Journal of Human Genetics, 2017, 62, 269-275.	2.3	18
29	Novel synonymous substitution in POMGNT1 promotes exon skipping in a patient with congenital muscular dystrophy. Journal of Human Genetics, 2008, 53, 565-572.	2.3	17
30	Bilirubin Dependence on UGT1A1 Polymorphisms, Hemoglobin, Fasting Time and Body Mass Index. American Journal of the Medical Sciences, 2012, 343, 114-118.	1.1	16
31	New massive parallel sequencing approach improves the genetic characterization of congenital myopathies. Journal of Human Genetics, 2016, 61, 497-505.	2.3	15
32	Identification of a novel AluSx-mediated deletion of exon 3 in the SBDS gene in a patient with Shwachmanâ€“Diamond syndrome. Blood Cells, Molecules, and Diseases, 2007, 39, 96-101.	1.4	14
33	Reviewing Large LAMA2 Deletions and Duplications in Congenital Muscular Dystrophy Patients. Journal of Neuromuscular Diseases, 2014, 1, 169-179.	2.6	14
34	Private dysferlin exon skipping mutation (c.5492G>A) with a founder effect reveals further alternative splicing involving exons 49â€“51. Journal of Human Genetics, 2010, 55, 546-549.	2.3	12
35	Glucose-6-phosphate dehydrogenase Aveiro: a de novo mutation associated with chronic nonspherocytic hemolytic anemia. Blood, 2000, 95, 1499-1501.	1.4	11
36	Identification of novel variants in ten patients with Hermansky-Pudlak syndrome by high-throughput sequencing. Annals of Medicine, 2019, 51, 141-148.	3.8	11

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37	A NEW CASE OF (TA)8 ALLELE IN THE UGT1A1 GENE PROMOTER IN A CAUCASIAN GIRL WITH GILBERTâ€™ SYNDROME. Pediatric Hematology and Oncology, 2004, 21, 371-374.	0.8	10
38	Hematologically important mutations: Shwachmanâ€“Diamond syndrome. Blood Cells, Molecules, and Diseases, 2008, 40, 183-184.	1.4	10
39	RYR1-Related Myopathies: Clinical, Histopathologic and Genetic Heterogeneity Among 17 Patients from a Portuguese Tertiary Centre. Journal of Neuromuscular Diseases, 2017, 4, 67-76.	2.6	10
40	Linkage disequilibrium between phenylketonuria and RFLP haplotype 1 at the phenylalanine hydroxylase locus in Portugal. Human Genetics, 1992, 89, 69-72.	3.8	9
41	Ryanodine Myopathies Without Central Coresâ€”Clinical, Histopathologic, and Genetic Description of Three Cases. Pediatric Neurology, 2014, 51, 275-278.	2.1	9
42	Novel ancestral Dysferlin splicing mutation which migrated from the Iberian peninsula to South America. Neuromuscular Disorders, 2011, 21, 328-337.	0.6	8
43	Î±IIbÎ²3 variants in ten families with autosomal dominant macrothrombocytopenia: Expanding the mutational and clinical spectrum. PLoS ONE, 2020, 15, e0235136.	2.5	8
44	Biochemical characterization of Î²-hexosaminidase in different biological specimens from eleven patients with GM2-gangliosidosis B1 variant. Journal of Inherited Metabolic Disease, 1991, 14, 715-720.	3.6	7
45	Variobox: Automatic Detection and Annotation of Human Genetic Variants. Human Mutation, 2014, 35, 202-207.	2.5	7
46	New Approaches in Molecular Diagnosis and Population Carrier Screening for Spinal Muscular Atrophy. Genetic Testing and Molecular Biomarkers, 2011, 15, 319-326.	0.7	6
47	FXTAS is rare among Portuguese patients with movement disorders: FMR1 premutations may be associated with a wider spectrum of phenotypes. Behavioral and Brain Functions, 2011, 7, 19.	3.3	6
48	Development and validation of a multiplex-PCR assay for X-linked intellectual disability. BMC Medical Genetics, 2013, 14, 80.	2.1	6
49	Classical fragile-X phenotype in a female infant disclosed by comprehensive genomic studies. BMC Medical Genetics, 2018, 19, 74.	2.1	6
50	Bilirubin Levels and Redox Status in a Young Healthy Population. Acta Haematologica, 2013, 130, 57-60.	1.4	4
51	A Portuguese case of Fukuyama congenital muscular dystrophy caused by a multi-exonic duplication in the fukutin gene. Neuromuscular Disorders, 2013, 23, 557-561.	0.6	4
52	Unveiling the genetic etiology of primary ciliary dyskinesia: When standard genetic approach is not enough. Advances in Medical Sciences, 2020, 65, 1-11.	2.1	4
53	Molecular Characterization of a Portuguese Patient with Shwachman-Diamond Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2005, 41, 115-116.	1.8	3
54	Identification of a novel deletion in UDP-glucuronosyltransferase gene in a patient with Criglerâ€“Najjar syndrome type I. Blood Cells, Molecules, and Diseases, 2009, 42, 265-266.	1.4	3

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55	A Family With 2 Different Hereditary Diseases Leading to Early Cardiac Involvement. Journal of Clinical Neuromuscular Disease, 2013, 14, 204-208.	0.7	3
56	A 26-Year Experience in Chorionic Villus Sampling Prenatal Genetic Diagnosis. Journal of Clinical Medicine, 2014, 3, 838-848.	2.4	3
57	Two Novel Pathogenic MID1 Variants and Genotype-Phenotype Correlation Reanalysis in X-Linked Opitz G/BBB Syndrome. Molecular Syndromology, 2018, 9, 45-51.	0.8	3
58	Homozygosity Mapping using Whole-Exome Sequencing: A Valuable Approach for Pathogenic Variant Identification in Genetic Diseases. , 2017, , .		3
59	Integrating Whole-Genome Sequencing in Clinical Genetics: A Novel Disruptive Structural Rearrangement Identified in the Dystrophin Gene (DMD). International Journal of Molecular Sciences, 2022, 23, 59.	4.1	3
60	Lipoprotein(a) Levels in Obese Portuguese Children and Adolescents: Contribution of the Pentanucleotide Repeat (TTTTA)n Polymorphism in the Apolipoprotein(a) Gene. JAMA Pediatrics, 2009, 163, 393.	3.0	2
61	Statistical Approach to Prenatal Zygosity Assessment Following a Decade of Molecular Aneuploidy Screening. Twin Research and Human Genetics, 2011, 14, 221-227.	0.6	2
62	Evaluating Runs of Homozygosity in Exome Sequencing Data - Utility in Disease Inheritance Model Selection and Variant Filtering. Communications in Computer and Information Science, 2018, , 268-288.	0.5	2
63	Development and Validation of a Mathematical Model to Predict the Complexity of FMR1 Allele Combinations. Frontiers in Genetics, 2020, 11, 557147.	2.3	2
64	Two Compound Heterozygous Variants in SNX14 Cause Stereotypies and Dystonia in Autosomal Recessive Spinocerebellar Ataxia 20. Frontiers in Genetics, 2020, 11, 1038.	2.3	2
65	Determination of neutrophil Fcgamma receptor IIIb antigens (HNA-1a, HNA-1b and HNA-1c) by fluorescence-primed allele-specific polymerase chain reaction. International Journal of Laboratory Hematology, 2004, 26, 319-321.	0.2	1
66	Development and validation in 500 female samples of a TP-PCR assay to identify AFF2 GCC expansions. Scientific Reports, 2021, 11, 14676.	3.3	1
67	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. , 0, .		1
68	CalDAG-GEFI Deficiency in a Family with Symptomatic Heterozygous and Homozygous Carriers of a Likely Pathogenic Variant in RASGRP2. International Journal of Molecular Sciences, 2021, 22, 12423.	4.1	1
69	Can the Synergic Contribution of Multigenic Variants Explain the Clinical and Cellular Phenotypes of a Neurodevelopmental Disorder?. Genes, 2022, 13, 78.	2.4	1
70	Single nucleotide polymorphisms in the apo(a) kringle IV type 8 domain are not associated with atherothrombotic serum lipoprotein (a) concentration, in a portuguese paediatric population. International Journal of Laboratory Hematology, 2008, 30, 240-243.	1.3	0
71	Haplotype analysis of newly diagnosed Portuguese and Brazilian families with fibrinogen amyloidosis caused by the FGA p.Glu545Val variant. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2019, 26, 144-145.	3.0	0
72	Usher syndrome and Nebulinâ€associated myopathy in a single patient due to variants in MYO7A and NEB. Clinical Case Reports (discontinued), 2020, 8, 2476-2482.	0.5	0

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73	A Rare Disease Patient Manager. Advances in Intelligent and Soft Computing, 2012, , 173-180.	0.2	0
74	Ten New Cases of Hermansky-Pudlak Syndrome in the Iberian Peninsula: Identification of Novel Genetic Variants in HPS3, HPS4, HPS6 and DTNBP1 Associated with Significant Clinical Complications. Blood, 2018, 132, 1147-1147.	1.4	0