Lourdes R Desviat

List of Publications by Year
in descending order

98 papers	3,101 citations	159525 30 h-index	197736 49 g-index
99 all docs	99 docs citations	99 times ranked	3298 citing authors

#	Article	IF	Citations
1	Delivery of oligonucleotideâ€based therapeutics: challenges and opportunities. EMBO Molecular Medicine, 2021, 13, e13243.	3.3	181
2	From The Cover: Correction of kinetic and stability defects by tetrahydrobiopterin in phenylketonuria patients with certain phenylalanine hydroxylase mutations. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 16903-16908.	3.3	156
3	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	2.6	138
4	Delivery is key: lessons learnt from developing spliceâ€switching antisense therapies. EMBO Molecular Medicine, 2017, 9, 545-557.	3.3	119
5	Phenylketonuria: Genotype-phenotype correlations based on expression analysis of structural and functional mutations inPAH. Human Mutation, 2003, 21, 370-378.	1.1	111
6	Mechanisms underlying responsiveness to tetrahydrobiopterin in mild phenylketonuria mutations. Human Mutation, 2004, 24, 388-399.	1.1	109
7	Spanish BH4-responsive phenylalanine hydroxylase-deficient patients: Evolution of seven patients on long-term treatment with tetrahydrobiopterin. Molecular Genetics and Metabolism, 2005, 86, 61-66.	0.5	109
8	Overview of mutations in thePCCA andPCCB genes causing propionic acidemia. , 1999, 14, 275-282.		81
9	Expression Analysis of Phenylketonuria Mutations. Journal of Biological Chemistry, 2000, 275, 29737-29742.	1.6	79
10	Genetic and cellular studies of oxidative stress in methylmalonic aciduria (MMA) cobalamin deficiency type C (cblC) with homocystinuria (MMACHC). Human Mutation, 2009, 30, 1558-1566.	1.1	76
11	The Molecular Basis of 3-Methylcrotonylglycinuria, a Disorder of Leucine Catabolism. American Journal of Human Genetics, 2001, 68, 334-346.	2.6	73
12	Genetic analysis of three genes causing isolated methylmalonic acidemia: identification of 21 novel allelic variants. Molecular Genetics and Metabolism, 2005, 84, 317-325.	0.5	67
13	A Novel Regulatory Defect in the Branched-Chain Î±-Keto Acid Dehydrogenase Complex Due to a Mutation in thePPM1KGene Causes a Mild Variant Phenotype of Maple Syrup Urine Disease. Human Mutation, 2013, 34, 355-362.	1.1	67
14	Tetrahydrobiopterin responsiveness: results of the BH4 loading test in 31 Spanish PKU patients and correlation with their genotype. Molecular Genetics and Metabolism, 2004, 83, 157-162.	0.5	56
15	Functional analysis of three splicing mutations identified in the PMM2 gene: Toward a new therapy for congenital disorder of glycosylation type Ia. Human Mutation, 2009, 30, 795-803.	1.1	46
16	Expression analysis revealing destabilizing mutations in phosphomannomutase 2 deficiency (PMM2â€CDG). Journal of Inherited Metabolic Disease, 2011, 34, 929-939.	1.7	45
17	Functional characterization of novel genotypes and cellular oxidative stress studies in propionic acidemia. Journal of Inherited Metabolic Disease, 2013, 36, 731-740.	1.7	44
18	Human Propionyl-CoA Carboxylase Î² Subunit Gene: Exon-Intron Definition and Mutation Spectrum in Spanish and Latin American Propionic Acidemia Patients. American Journal of Human Genetics, 1998, 63, 360-369.	2.6	43

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19	A Novel Dominant Hyperekplexia Mutation Y705C Alters Trafficking and Biochemical Properties of the Presynaptic Glycine Transporter GlyT2. Journal of Biological Chemistry, 2012, 287, 28986-29002.	1.6	42
20	Therapeutic strategies based on modified U1 snRNAs and chaperones for Sanfilippo C splicing mutations. Orphanet Journal of Rare Diseases, 2014, 9, 180.	1.2	42
21	Kinetic and stability analysis of PKU mutations identified in BH4-responsive patients. Molecular Genetics and Metabolism, 2005, 86, 11-16.	0.5	38
22	Present and future of antisense therapy for splicing modulation in inherited metabolic disease. Journal of Inherited Metabolic Disease, 2010, 33, 397-403.	1.7	38
23	The Effects of PMM2-CDG-Causing Mutations on the Folding, Activity, and Stability of the PMM2 Protein. Human Mutation, 2015, 36, 851-860.	1.1	38
24	Structure of the PCCA Gene and Distribution of Mutations Causing Propionic Acidemia. Molecular Genetics and Metabolism, 2001, 74, 238-247.	0.5	37
25	Quantitative Analysis of Mitochondrial Protein Expression in Methylmalonic Acidemia by Two-Dimensional Difference Gel Electrophoresis. Journal of Proteome Research, 2006, 5, 1602-1610.	1.8	37
26	Antioxidants successfully reduce ROS production in propionic acidemia fibroblasts. Biochemical and Biophysical Research Communications, 2014, 452, 457-461.	1.0	37
27	Pharmacological Chaperoning: A Potential Treatment for PMM2-CDG. Human Mutation, 2017, 38, 160-168.	1.1	37
28	Functional characterization of PCCA mutations causing propionic acidemia. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 119-125.	1.8	36
29	Accurate molecular diagnosis of phenylketonuria and tetrahydrobiopterin-deficient hyperphenylalaninemias using high-throughput targeted sequencing. European Journal of Human Genetics, 2014, 22, 528-534.	1.4	36
30	Pharmacological chaperones as a potential therapeutic option in methylmalonic aciduria cblB type. Human Molecular Genetics, 2013, 22, 3680-3689.	1.4	33
31	Molecular diagnosis of glycogen storage disease and disorders with overlapping clinical symptoms by massive parallel sequencing. Genetics in Medicine, 2016, 18, 1037-1043.	1.1	32
32	Oxidative stress and apoptosis in homocystinuria patients with genetic remethylation defects. Journal of Cellular Biochemistry, 2013, 114, 183-191.	1.2	31
33	Analysis of the effect of tetrahydrobiopterin on PAH gene expression in hepatoma cells. FEBS Letters, 2006, 580, 1697-1701.	1.3	30
34	Genotypeâ€“phenotype correlations in sepiapterin reductase deficiency. A splicing defect accounts for a new phenotypic variant. Neurogenetics, 2011, 12, 183-191.	0.7	30
35	Role of miRNAs in human disease and inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2017, 40, 471-480.	1.7	30
36	Three novel splice mutations in the PCCA gene causing identical exon skipping in propionic acidemia patients. Human Genetics, 1997, 101, 93-96.	1.8	29

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37	High frequency of large genomic deletions in the PCCA gene causing propionic acidemia. Molecular Genetics and Metabolism, 2009, 96, 171-176.	0.5	29
38	Treatment with antioxidants ameliorates oxidative damage in a mouse model of propionic acidemia. Molecular Genetics and Metabolism, 2017, 122, 43-50.	0.5	29
39	Minigenes to Confirm Exon Skipping Mutations. Methods in Molecular Biology, 2012, 867, 37-47.	0.4	28
40	45-Year-old female with propionic acidemia, renal failure, and premature ovarian failure; late complications of propionic acidemia?. Molecular Genetics and Metabolism, 2011, 103, 338-340.	0.5	27
41	Feasibility of nonsense mutation readthrough as a novel therapeutical approach in propionic acidemia. Human Mutation, 2012, 33, 973-980.	1.1	26
42	Molecular epidemiology, genotypeâ€“phenotype correlation and BH4 responsiveness in Spanish patients with phenylketonuria. Journal of Human Genetics, 2016, 61, 731-744.	1.1	26
43	Intronic PAH gene mutations cause a splicing defect by a novel mechanism involving U1snRNP binding downstream of the 5â€™ splice site. PLoS Genetics, 2018, 14, e1007360.	1.5	26
44	Pseudoexon exclusion by antisense therapy in 6-pyruvoyl-tetrahydropterin synthase deficiency. Human Mutation, 2011, 32, 1019-1027.	1.1	25
45	Altered Redox Homeostasis in Branched-Chain Amino Acid Disorders, Organic Acidurias, and Homocystinuria. Oxidative Medicine and Cellular Longevity, 2018, 2018, 1-17.	1.9	24
46	Functional and structural analysis of five mutations identified in methylmalonic aciduria cblB type. Human Mutation, 2010, 31, 1033-1042.	1.1	23
47	Value of genetic analysis for confirming inborn errors of metabolism detected through the Spanish neonatal screening program. European Journal of Human Genetics, 2019, 27, 556-562.	1.4	23
48	Pathogenic variants of DNAJC12 and evaluation of the encoded cochaperone as a genetic modifier of hyperphenylalaninemia. Human Mutation, 2020, 41, 1329-1338.	1.1	23
49	Genetic heterogeneity in propionic acidemia patients with Î±-subunit defects. Identification of five novel mutations, one of them causing instability of the protein. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1999, 1453, 351-358.	1.8	22
50	New splicing mutations in propionic acidemia. Journal of Human Genetics, 2006, 51, 992-997.	1.1	22
51	DPAGT1-CDG: Functional analysis of disease-causing pathogenic mutations and role of endoplasmic reticulum stress. PLoS ONE, 2017, 12, e0179456.	1.1	22
52	Phenylketonuria in Spain: RFLP haplotypes and linked mutations. Human Genetics, 1993, 92, 254-8.	1.8	20
53	Identification of exonic deletions in the PAH gene causing phenylketonuria by MLPA analysis. Clinica Chimica Acta, 2006, 373, 164-167.	0.5	20
54	Molecular epidemiology and genotypeâ€“phenotype correlation in phenylketonuria patients from South Spain. Journal of Human Genetics, 2013, 58, 279-284.	1.1	20

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55	Endoplasmic Reticulum Stress and Autophagy in Homocystinuria Patients with Remethylation Defects. PLoS ONE, 2016, 11, e0150357.	1.1	20
56	Identification of 34 novel mutations in propionic acidemia: Functional characterization of missense variants and phenotype associations. Molecular Genetics and Metabolism, 2018, 125, 266-275.	0.5	19
57	Overexpression of adapted U1snRNA in patients' cells to correct a 5â€² splice site mutation in propionic acidemia. Molecular Genetics and Metabolism, 2011, 102, 134-138.	0.5	18
58	Presence of the Mediterranean PKU mutation IVS10 in Latin America. Human Molecular Genetics, 1993, 2, 1289-1290.	1.4	17
59	The STR252 - IVS10nt546 - VNTR7 phenylalanine hydroxylase minihaplotype in five Mediterranean samples. Human Genetics, 1997, 100, 350-355.	1.8	17
60	Qualitative and quantitative analysis of the effect of splicing mutations in propionic acidemia underlying non-severe phenotypes. Human Genetics, 2004, 115, 239-47.	1.8	17
61	The activity of wild type and mutant phenylalanine hydroxylase with respect to the C-oxidation of phenylalanine and the S-oxidation of S-carboxymethyl-l-cysteine. Molecular Genetics and Metabolism, 2009, 96, 27-31.	0.5	17
62	Cardiac Complications of Propionic and Other Inherited Organic Acidemias. Frontiers in Cardiovascular Medicine, 2020, 7, 617451.	1.1	17
63	Mutation analysis of phenylketonuria patients from Morocco: High prevalence of mutation G352fsdelG and detection of a novel mutation p.K85X. Clinical Biochemistry, 2010, 43, 76-81.	0.8	16
64	Understanding molecular mechanisms in propionic acidemia and investigated therapeutic strategies. Expert Opinion on Orphan Drugs, 2015, 3, 1427-1438.	0.5	16
65	Dysregulated miRNAs and their pathogenic implications for the neurometabolic disease propionic acidemia. Scientific Reports, 2017, 7, 5727.	1.6	16
66	Antisense Mediated Splicing Modulation For Inherited Metabolic Diseases: Challenges for Delivery. Nucleic Acid Therapeutics, 2014, 24, 48-56.	2.0	15
67	The molecular landscape of propionic acidemia and methylmalonic aciduria in Latin America. Journal of Inherited Metabolic Disease, 2010, 33, 307-314.	1.7	14
68	Novel features in the evolution of adenylosuccinate lyase deficiency. European Journal of Paediatric Neurology, 2012, 16, 343-348.	0.7	14
69	Generation and characterization of a human iPSC line from a patient with propionic acidemia due to defects in the PCCA gene. Stem Cell Research, 2017, 23, 173-177.	0.3	14
70	Identification of novel mutations in the PCCB gene in European propionic acidemia patients. , 1999, 14, 89-90.		13
71	Functional analysis of splicing mutations in the IDS gene and the use of antisense oligonucleotides to exploit an alternative therapy for MPS II. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 2712-2721.	1.8	13
72	Genes and Variants Underlying Human Congenital Lactic Acidosisâ€"From Genetics to Personalized Treatment. Journal of Clinical Medicine, 2019, 8, 1811.	1.0	13

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73	BH4 responsiveness associated to a PKU mutation with decreased binding affinity for the cofactor. Clinica Chimica Acta, 2007, 380, 8-12.	0.5	11
74	Segmental uniparental disomy leading to homozygosity for a pathogenic mutation in three recessive metabolic diseases. Molecular Genetics and Metabolism, 2012, 105, 270-271.	0.5	11
75	Pathogenic implications of dysregulated miRNAs in propionic acidemia related cardiomyopathy. Translational Research, 2020, 218, 43-56.	2.2	11
76	Mutation analysis of phenylketonuria in South Brazil. Human Mutation, 1996, 8, 262-264.	1.1	10
77	Phenylketonuria in Spanish Gypsies: Prevalence of the IVS10nt546 mutation on haplotype 34. Human Mutation, 1997, 9, 66-68.	1.1	10
78	Towards a model to explain the intragenic complementation in the heteromultimeric protein propionyl-CoA carboxylase. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2005, 1740, 489-498.	1.8	10
79	Molecular basis of phenylketonuria in Venezuela: Presence of two novel null mutations. Human Mutation, 1998, 11, 354-359.	1.1	9
80	Expression analysis of mutation P244L, which causes mild hyperphenylalaninemia. Human Mutation, 1995, 5, 188-190.	1.1	8
81	The Missense p.S231F Phenylalanine Hydroxylase Gene Mutation Causes Complete Loss of Enzymatic Activity In Vitro. Protein Journal, 2009, 28, 294-299.	0.7	8
82	A Sensitive Assay System To Test Antisense Oligonucleotides for Splice Suppression Therapy in the Mouse Liver. Molecular Therapy - Nucleic Acids, 2014, 3, e193.	2.3	7
83	Antisense Oligonucleotide Rescue of Deep-Intronic Variants Activating Pseudoexons in the 6-Pyruvoyl-Tetrahydropterin Synthase Gene. Nucleic Acid Therapeutics, 2022, 32, 378-390.	2.0	7
84	Identification of the first deletionâ€“insertion involving the complete structure of GAA gene and part of CCDC40 gene mediated by an Alu element. Gene, 2013, 519, 169-172.	1.0	6
85	Data in support of a functional analysis of splicing mutations in the IDS gene and the use of antisense oligonucleotides to exploit an alternative therapy for MPS II. Data in Brief, 2015, 5, 810-817.	0.5	6
86	RNA solutions to treat inborn errors of metabolism. Molecular Genetics and Metabolism, 2022, 136, 289-295.	0.5	6
87	Analysis of Defective Subunit Interactions Using the Two-Hybrid System. , 2003, 232, 245-256.		4
88	Generation and characterization of a human iPSC line (UAMi004-A) from a patient with propionic acidemia due to defects in the PCCB gene. Stem Cell Research, 2019, 38, 101469.	0.3	4
89	Isolated and Combined Remethylation Disorders. FIRE Forum for International Research in Education, 2017, 5, 232640981668573.	0.7	3
90	Improving the diagnosis of cobalamin and related defects by genomic analysis, plus functional and structural assessment of novel variants. Orphanet Journal of Rare Diseases, 2018, 13, 125.	1.2	3

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91	Molecular characterization of phenylalanine hydroxylase deficiency in Chile. Human Mutation, 1999, 13, 503-503.	1.1	2
92	Tetrahydrobiopterin deficiency among Serbian patients presenting with hyperphenylalaninemia. Journal of Pediatric Endocrinology and Metabolism, 2014, 28, 477-80.	0.4	2
93	Functional Characterization of Novel Phenylalanine Hydroxylase p.Gln226Lys Mutation Revealed Its Non-responsiveness to Tetrahydrobiopterin Treatment in Hepatoma Cellular Model. Biochemical Genetics, 2018, 56, 533-541.	0.8	2
94	COST Actions: fostering collaborative research for rare diseases. Lancet Neurology, The, 2019, 18, 989-991.	4.9	2
95	Generation of a gene-corrected human isogenic line (UAMi006-A) from propionic acidemia patient iPSC with an homozygous mutation in the PCCB gene using CRISPR/Cas9 technology. Stem Cell Research, 2020, 49, 102055.	0.3	2
96	Functional analysis of a novel mutation in the PCCA gene identified in a late-infantile onset propionic acidemia patient. Clinica Chimica Acta, 2010, 411, 1388-1389.	0.5	1
97	RNA-Based Therapies for Inherited Metabolic Diseases. , 2012, , 357-370.		1
98	Modeling Splicing Variants Amenable to Antisense Therapy by Use of CRISPR-Cas9-Based Gene Editing in HepG2 Cells. Methods in Molecular Biology, 2022, 2434, 167-184.	0.4	1