

Lourdes R Desviat

List of Publications by Year in descending order

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#	ARTICLE	IF	CITATIONS
1	Modeling Splicing Variants Amenable to Antisense Therapy by Use of CRISPR-Cas9-Based Gene Editing in HepG2 Cells. <i>Methods in Molecular Biology</i> , 2022, 2434, 167-184.	0.4	1
2	Antisense Oligonucleotide Rescue of Deep-Intronic Variants Activating Pseudoexons in the 6-Pyruvoyl-Tetrahydropterin Synthase Gene. <i>Nucleic Acid Therapeutics</i> , 2022, 32, 378-390.	2.0	7
3	RNA solutions to treat inborn errors of metabolism. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 289-295.	0.5	6
4	Delivery of oligonucleotide-based therapeutics: challenges and opportunities. <i>EMBO Molecular Medicine</i> , 2021, 13, e13243.	3.3	181
5	The Genetic Landscape and Epidemiology of Phenylketonuria. <i>American Journal of Human Genetics</i> , 2020, 107, 234-250.	2.6	138
6	Cardiac Complications of Propionic and Other Inherited Organic Acidemias. <i>Frontiers in Cardiovascular Medicine</i> , 2020, 7, 617451.	1.1	17
7	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. <i>Stem Cell Research</i> , 2020, 49, 102055.	0.3	2
8	Pathogenic implications of dysregulated miRNAs in propionic acidemia related cardiomyopathy. <i>Translational Research</i> , 2020, 218, 43-56.	2.2	11
9	Pathogenic variants of <i>DNAJC12</i> and evaluation of the encoded cochaperone as a genetic modifier of hyperphenylalaninemia. <i>Human Mutation</i> , 2020, 41, 1329-1338.	1.1	23
10	COST Actions: fostering collaborative research for rare diseases. <i>Lancet Neurology</i> , The, 2019, 18, 989-991.	4.9	2
11	Genes and Variants Underlying Human Congenital Lactic Acidosis—From Genetics to Personalized Treatment. <i>Journal of Clinical Medicine</i> , 2019, 8, 1811.	1.0	13
12	Generation and characterization of a human iPSC line (UAMi004-A) from a patient with propionic acidemia due to defects in the PCCB gene. <i>Stem Cell Research</i> , 2019, 38, 101469.	0.3	4
13	Value of genetic analysis for confirming inborn errors of metabolism detected through the Spanish neonatal screening program. <i>European Journal of Human Genetics</i> , 2019, 27, 556-562.	1.4	23
14	Functional Characterization of Novel Phenylalanine Hydroxylase p.Gln226Lys Mutation Revealed Its Non-responsiveness to Tetrahydrobiopterin Treatment in Hepatoma Cellular Model. <i>Biochemical Genetics</i> , 2018, 56, 533-541.	0.8	2
15	Identification of 34 novel mutations in propionic acidemia: Functional characterization of missense variants and phenotype associations. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 266-275.	0.5	19
16	Altered Redox Homeostasis in Branched-Chain Amino Acid Disorders, Organic Acidurias, and Homocystinuria. <i>Oxidative Medicine and Cellular Longevity</i> , 2018, 2018, 1-17.	1.9	24
17	Improving the diagnosis of cobalamin and related defects by genomic analysis, plus functional and structural assessment of novel variants. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 125.	1.2	3
18	Intronic PAH gene mutations cause a splicing defect by a novel mechanism involving U1snRNP binding downstream of the 5' splice site. <i>PLoS Genetics</i> , 2018, 14, e1007360.	1.5	26

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19	Role of miRNAs in human disease and inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 471-480.	1.7	30
20	Isolated and Combined Remethylation Disorders. <i>FIRE Forum for International Research in Education</i> , 2017, 5, 232640981668573.	0.7	3
21	Delivery is key: lessons learnt from developing splice-switching antisense therapies. <i>EMBO Molecular Medicine</i> , 2017, 9, 545-557.	3.3	119
22	Treatment with antioxidants ameliorates oxidative damage in a mouse model of propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 43-50.	0.5	29
23	Dysregulated miRNAs and their pathogenic implications for the neurometabolic disease propionic acidemia. <i>Scientific Reports</i> , 2017, 7, 5727.	1.6	16
24	Generation and characterization of a human iPSC line from a patient with propionic acidemia due to defects in the PCCA gene. <i>Stem Cell Research</i> , 2017, 23, 173-177.	0.3	14
25	Pharmacological Chaperoning: A Potential Treatment for PMM2-CDG. <i>Human Mutation</i> , 2017, 38, 160-168.	1.1	37
26	DPAGT1-CDG: Functional analysis of disease-causing pathogenic mutations and role of endoplasmic reticulum stress. <i>PLoS ONE</i> , 2017, 12, e0179456.	1.1	22
27	Molecular epidemiology, genotype-phenotype correlation and BH4 responsiveness in Spanish patients with phenylketonuria. <i>Journal of Human Genetics</i> , 2016, 61, 731-744.	1.1	26
28	Molecular diagnosis of glycogen storage disease and disorders with overlapping clinical symptoms by massive parallel sequencing. <i>Genetics in Medicine</i> , 2016, 18, 1037-1043.	1.1	32
29	Endoplasmic Reticulum Stress and Autophagy in Homocystinuria Patients with Remethylation Defects. <i>PLoS ONE</i> , 2016, 11, e0150357.	1.1	20
30	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. <i>Data in Brief</i> , 2015, 5, 810-817.	0.5	6
31	The Effects of PMM2-CDG-Causing Mutations on the Folding, Activity, and Stability of the PMM2 Protein. <i>Human Mutation</i> , 2015, 36, 851-860.	1.1	38
32	Understanding molecular mechanisms in propionic acidemia and investigated therapeutic strategies. <i>Expert Opinion on Orphan Drugs</i> , 2015, 3, 1427-1438.	0.5	16
33	Functional analysis of splicing mutations in the IDS gene and the use of antisense oligonucleotides to exploit an alternative therapy for MPS II. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2015, 1852, 2712-2721.	1.8	13
34	Therapeutic strategies based on modified U1 snRNAs and chaperones for Sanfilippo C splicing mutations. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 180.	1.2	42
35	A Sensitive Assay System To Test Antisense Oligonucleotides for Splice Suppression Therapy in the Mouse Liver. <i>Molecular Therapy - Nucleic Acids</i> , 2014, 3, e193.	2.3	7
36	Tetrahydrobiopterin deficiency among Serbian patients presenting with hyperphenylalaninemia. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014, 28, 477-80.	0.4	2

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37	Antisense Mediated Splicing Modulation For Inherited Metabolic Diseases: Challenges for Delivery. <i>Nucleic Acid Therapeutics</i> , 2014, 24, 48-56.	2.0	15
38	Accurate molecular diagnosis of phenylketonuria and tetrahydrobiopterin-deficient hyperphenylalaninemias using high-throughput targeted sequencing. <i>European Journal of Human Genetics</i> , 2014, 22, 528-534.	1.4	36
39	Antioxidants successfully reduce ROS production in propionic acidemia fibroblasts. <i>Biochemical and Biophysical Research Communications</i> , 2014, 452, 457-461.	1.0	37
40	A Novel Regulatory Defect in the Branched-Chain α -Keto Acid Dehydrogenase Complex Due to a Mutation in the <i>PPM1K</i> Gene Causes a Mild Variant Phenotype of Maple Syrup Urine Disease. <i>Human Mutation</i> , 2013, 34, 355-362.	1.1	67
41	Oxidative stress and apoptosis in homocystinuria patients with genetic remethylation defects. <i>Journal of Cellular Biochemistry</i> , 2013, 114, 183-191.	1.2	31
42	Identification of the first deletion-insertion involving the complete structure of GAA gene and part of CCDC40 gene mediated by an Alu element. <i>Gene</i> , 2013, 519, 169-172.	1.0	6
43	Functional characterization of novel genotypes and cellular oxidative stress studies in propionic acidemia. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 731-740.	1.7	44
44	Pharmacological chaperones as a potential therapeutic option in methylmalonic aciduria cblB type. <i>Human Molecular Genetics</i> , 2013, 22, 3680-3689.	1.4	33
45	Molecular epidemiology and genotype-phenotype correlation in phenylketonuria patients from South Spain. <i>Journal of Human Genetics</i> , 2013, 58, 279-284.	1.1	20
46	A Novel Dominant Hyperekplexia Mutation Y705C Alters Trafficking and Biochemical Properties of the Presynaptic Glycine Transporter GlyT2. <i>Journal of Biological Chemistry</i> , 2012, 287, 28986-29002.	1.6	42
47	Segmental uniparental disomy leading to homozygosity for a pathogenic mutation in three recessive metabolic diseases. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 270-271.	0.5	11
48	RNA-Based Therapies for Inherited Metabolic Diseases. , 2012, , 357-370.		1
49	Minigenes to Confirm Exon Skipping Mutations. <i>Methods in Molecular Biology</i> , 2012, 867, 37-47.	0.4	28
50	Feasibility of nonsense mutation readthrough as a novel therapeutical approach in propionic acidemia. <i>Human Mutation</i> , 2012, 33, 973-980.	1.1	26
51	Novel features in the evolution of adenylosuccinate lyase deficiency. <i>European Journal of Paediatric Neurology</i> , 2012, 16, 343-348.	0.7	14
52	Overexpression of adapted U1snRNA in patients' cells to correct a 5' splice site mutation in propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2011, 102, 134-138.	0.5	18
53	45-Year-old female with propionic acidemia, renal failure, and premature ovarian failure; late complications of propionic acidemia?. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 338-340.	0.5	27
54	Expression analysis revealing destabilizing mutations in phosphomannomutase 2 deficiency (PMM2-CDG). <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 929-939.	1.7	45

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55	Genotype-phenotype correlations in sepiapterin reductase deficiency. A splicing defect accounts for a new phenotypic variant. <i>Neurogenetics</i> , 2011, 12, 183-191.	0.7	30
56	Pseudoexon exclusion by antisense therapy in 6-pyruvoyl-tetrahydropterin synthase deficiency. <i>Human Mutation</i> , 2011, 32, 1019-1027.	1.1	25
57	The molecular landscape of propionic acidemia and methylmalonic aciduria in Latin America. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 307-314.	1.7	14
58	Present and future of antisense therapy for splicing modulation in inherited metabolic disease. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 397-403.	1.7	38
59	Functional and structural analysis of five mutations identified in methylmalonic aciduria cblB type. <i>Human Mutation</i> , 2010, 31, 1033-1042.	1.1	23
60	Mutation analysis of phenylketonuria patients from Morocco: High prevalence of mutation G352fsdelG and detection of a novel mutation p.K85X. <i>Clinical Biochemistry</i> , 2010, 43, 76-81.	0.8	16
61	Functional analysis of a novel mutation in the PCCA gene identified in a late-infantile onset propionic acidemia patient. <i>Clinica Chimica Acta</i> , 2010, 411, 1388-1389.	0.5	1
62	Functional analysis of three splicing mutations identified in the PMM2 gene: Toward a new therapy for congenital disorder of glycosylation type Ia. <i>Human Mutation</i> , 2009, 30, 795-803.	1.1	46
63	Genetic and cellular studies of oxidative stress in methylmalonic aciduria (MMA) cobalamin deficiency type C (cblC) with homocystinuria (MMACHC). <i>Human Mutation</i> , 2009, 30, 1558-1566.	1.1	76
64	The Missense p.S231F Phenylalanine Hydroxylase Gene Mutation Causes Complete Loss of Enzymatic Activity In Vitro. <i>Protein Journal</i> , 2009, 28, 294-299.	0.7	8
65	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. <i>Molecular Genetics and Metabolism</i> , 2009, 96, 27-31.	0.5	17
66	High frequency of large genomic deletions in the PCCA gene causing propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2009, 96, 171-176.	0.5	29
67	BH4 responsiveness associated to a PKU mutation with decreased binding affinity for the cofactor. <i>Clinica Chimica Acta</i> , 2007, 380, 8-12.	0.5	11
68	Quantitative Analysis of Mitochondrial Protein Expression in Methylmalonic Acidemia by Two-Dimensional Difference Gel Electrophoresis. <i>Journal of Proteome Research</i> , 2006, 5, 1602-1610.	1.8	37
69	Analysis of the effect of tetrahydrobiopterin on PAH gene expression in hepatoma cells. <i>FEBS Letters</i> , 2006, 580, 1697-1701.	1.3	30
70	Identification of exonic deletions in the PAH gene causing phenylketonuria by MLPA analysis. <i>Clinica Chimica Acta</i> , 2006, 373, 164-167.	0.5	20
71	New splicing mutations in propionic acidemia. <i>Journal of Human Genetics</i> , 2006, 51, 992-997.	1.1	22
72	Towards a model to explain the intragenic complementation in the heteromultimeric protein propionyl-CoA carboxylase. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2005, 1740, 489-498.	1.8	10

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73	Genetic analysis of three genes causing isolated methylmalonic acidemia: identification of 21 novel allelic variants. <i>Molecular Genetics and Metabolism</i> , 2005, 84, 317-325.	0.5	67
74	Kinetic and stability analysis of PKU mutations identified in BH4-responsive patients. <i>Molecular Genetics and Metabolism</i> , 2005, 86, 11-16.	0.5	38
75	Spanish BH4-responsive phenylalanine hydroxylase-deficient patients: Evolution of seven patients on long-term treatment with tetrahydrobiopterin. <i>Molecular Genetics and Metabolism</i> , 2005, 86, 61-66.	0.5	109
76	From The Cover: Correction of kinetic and stability defects by tetrahydrobiopterin in phenylketonuria patients with certain phenylalanine hydroxylase mutations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 16903-16908.	3.3	156
77	Qualitative and quantitative analysis of the effect of splicing mutations in propionic acidemia underlying non-severe phenotypes. <i>Human Genetics</i> , 2004, 115, 239-47.	1.8	17
78	Mechanisms underlying responsiveness to tetrahydrobiopterin in mild phenylketonuria mutations. <i>Human Mutation</i> , 2004, 24, 388-399.	1.1	109
79	Tetrahydrobiopterin responsiveness: results of the BH4 loading test in 31 Spanish PKU patients and correlation with their genotype. <i>Molecular Genetics and Metabolism</i> , 2004, 83, 157-162.	0.5	56
80	Phenylketonuria: Genotype-phenotype correlations based on expression analysis of structural and functional mutations in PAH. <i>Human Mutation</i> , 2003, 21, 370-378.	1.1	111
81	Analysis of Defective Subunit Interactions Using the Two-Hybrid System. , 2003, 232, 245-256.		4
82	Functional characterization of PCCA mutations causing propionic acidemia. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2002, 1588, 119-125.	1.8	36
83	Structure of the PCCA Gene and Distribution of Mutations Causing Propionic Acidemia. <i>Molecular Genetics and Metabolism</i> , 2001, 74, 238-247.	0.5	37
84	The Molecular Basis of 3-Methylcrotonylglycinuria, a Disorder of Leucine Catabolism. <i>American Journal of Human Genetics</i> , 2001, 68, 334-346.	2.6	73
85	Expression Analysis of Phenylketonuria Mutations. <i>Journal of Biological Chemistry</i> , 2000, 275, 29737-29742.	1.6	79
86	Molecular characterization of phenylalanine hydroxylase deficiency in Chile. <i>Human Mutation</i> , 1999, 13, 503-503.	1.1	2
87	Identification of novel mutations in the PCCB gene in European propionic acidemia patients. , 1999, 14, 89-90.		13
88	Overview of mutations in the PCCA and PCCB genes causing propionic acidemia. , 1999, 14, 275-282.		81
89	Genetic heterogeneity in propionic acidemia patients with β -subunit defects. Identification of five novel mutations, one of them causing instability of the protein. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1999, 1453, 351-358.	1.8	22
90	Molecular basis of phenylketonuria in Venezuela: Presence of two novel null mutations. <i>Human Mutation</i> , 1998, 11, 354-359.	1.1	9

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91	Human Propionyl-CoA Carboxylase β Subunit Gene: Exon-Intron Definition and Mutation Spectrum in Spanish and Latin American Propionic Acidemia Patients. <i>American Journal of Human Genetics</i> , 1998, 63, 360-369.	2.6	43
92	The STR252 - IVS10nt546 - VNTR7 phenylalanine hydroxylase minihaplotype in five Mediterranean samples. <i>Human Genetics</i> , 1997, 100, 350-355.	1.8	17
93	Three novel splice mutations in the PCCA gene causing identical exon skipping in propionic acidemia patients. <i>Human Genetics</i> , 1997, 101, 93-96.	1.8	29
94	Phenylketonuria in Spanish Gypsies: Prevalence of the IVS10nt546 mutation on haplotype 34. <i>Human Mutation</i> , 1997, 9, 66-68.	1.1	10
95	Mutation analysis of phenylketonuria in South Brazil. <i>Human Mutation</i> , 1996, 8, 262-264.	1.1	10
96	Expression analysis of mutation P244L, which causes mild hyperphenylalaninemia. <i>Human Mutation</i> , 1995, 5, 188-190.	1.1	8
97	Phenylketonuria in Spain: RFLP haplotypes and linked mutations. <i>Human Genetics</i> , 1993, 92, 254-8.	1.8	20
98	Presence of the Mediterranean PKU mutation IVS10 in Latin America. <i>Human Molecular Genetics</i> , 1993, 2, 1289-1290.	1.4	17