Guillem Pintos-Morell

List of Publications by Year
in descending order

52 papers	2,328 citations	331670 21 h-index	214800 47 g-index
56 all docs	56 docs citations	56 times ranked	3209 citing authors

#	Article	IF	Citations
1	Suggested guidelines for the diagnosis and management of urea cycle disorders: First revision. Journal of Inherited Metabolic Disease, 2019, 42, 1192-1230.	3.6	277
2	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€"dystonia. Nature Communications, 2016, 7, 11601.	12.8	233
3	Clinical manifestations of Fabry disease in children: Data from the Fabry Outcome Survey. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 86-92.	1.5	184
4	Recommendations for the management of tyrosinaemia type 1. Orphanet Journal of Rare Diseases, 2013, 8, 8.	2.7	182
5	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	3.6	175
6	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2015, 138, 2847-2858.	7.6	128
7	Nephrin mutations cause childhood- and adult-onset focal segmental glomerulosclerosis. Kidney International, 2009, 76, 1268-1276.	5.2	111
8	Effects of enzyme replacement therapy in Fabry diseaseâ€"A comprehensive review of the medical literature. Genetics in Medicine, 2010, 12, 668-679.	2.4	100
9	TRPC6 mutational analysis in a large cohort of patients with focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2009, 24, 3089-3096.	0.7	99
10	Long-term effectiveness of agalsidase alfa enzyme replacement in Fabry disease: A Fabry Outcome Survey analysis. Molecular Genetics and Metabolism Reports, 2015, 3, 21-27.	1.1	97
11	Urinary biomarker investigation in children with Fabry disease using tandem mass spectrometry. Clinica Chimica Acta, 2015, 438, 195-204.	1.1	62
12	Genotypic and phenotypic features of all Spanish patients with McArdle disease: a 2016 update. BMC Genomics, 2017, 18, 819.	2.8	53
13	Usefulness of tissue Doppler on early detection of cardiac disease in Fabry patients and potential role of enzyme replacement therapy (ERT) for avoiding progression of disease. European Journal of Echocardiography, 2011, 12, 671-677.	2.3	49
14	Fabry disease in children and the effects of enzyme replacement treatment. European Journal of Pediatrics, 2009, 168, 1355-1363.	2.7	48
15	Clinical usefulness of tissue Doppler imaging in predicting preclinical Fabry cardiomyopathy. International Journal of Cardiology, 2009, 132, 38-44.	1.7	45
16	Tyrosinemia type 1 in Spain: Mutational analysis, treatment and longâ€term outcome. Pediatrics International, 2011, 53, 985-989.	0.5	37
17	Urea cycle disorders in Spain: an observational, cross-sectional and multicentric study of 104 cases. Orphanet Journal of Rare Diseases, 2014, 9, 187.	2.7	34
18	Therapeutic goals in the treatment of Fabry disease. Genetics in Medicine, 2010, 12, 713-720.	2.4	33

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19	Measuring patient experiences in Fabry disease: validation of the Fabry-specific Pediatric Health and Pain Questionnaire (FPHPQ). Health and Quality of Life Outcomes, 2012, 10, 116.	2.4	33
20	Rare Neurodegenerative Diseases: Clinical and Genetic Update. Advances in Experimental Medicine and Biology, 2017, 1031, 443-496.	1.6	30
21	Analysis of Renal and Cardiac Outcomes in Male Participants in the Fabry Outcome Survey Starting Agalsidase Alfa Enzyme Replacement Therapy Before and After 18 Years of Age . Drug Design, Development and Therapy, 2020, Volume 14, 2149-2158.	4.3	25
22	A DM1 family with interruptions associated with atypical symptoms and late onset but not with a milder phenotype. Human Mutation, 2020, 41, 420-431.	2.5	24
23	Exercise and Preexercise Nutrition as Treatment for McArdle Disease. Medicine and Science in Sports and Exercise, 2016, 48, 673-679.	0.4	20
24	Cardio- Renal Outcomes With Long- Term Agalsidase Alfa Enzyme Replacement Therapy: A 10-Year Fabry Outcome Survey (FOS) Analysis . Drug Design, Development and Therapy, 2019, Volume 13, 3705-3715.	4.3	19
25	Molecular analysis of mucopolysaccharidosis IVA (Morquio A) in Spain. Molecular Genetics and Metabolism, 2012, 106, 196-201.	1.1	18
26	Paediatric Fabry disease: prognostic significance of ocular changes for disease severity. BMC Ophthalmology, 2016, 16, 202.	1.4	18
27	A novel nonstop mutation in TYMP does not induce nonstop mRNA decay in a MNGIE patient with severe neuropathy. Human Mutation, 2011, 32, E2061-8.	2.5	15
28	Genes and exercise intolerance: insights from McArdle disease. Physiological Genomics, 2016, 48, 93-100.	2.3	15
29	Twenty years of the Fabry Outcome Survey (FOS): insights, achievements, and lessons learned from a global patient registry. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	14
30	Long-term follow-up of a patient with primary hypomagnesaemia and secondary hypocalcaemia due to a novel TRPM6 mutation. European Journal of Pediatrics, 2009, 168, 439-442.	2.7	13
31	TransiciÃ³n coordinada del paciente con cistinosis desde la medicina pediÃ¡trica a la medicina del adulto. Nefrologia, 2016, 36, 616-630.	0.4	13
32	Missense mutations have unexpected consequences: The McArdle disease paradigm. Human Mutation, 2018, 39, 1338-1343.	2.5	13
33	Nanotechnologyâ€based approaches for treating lysosomal storage disorders, a focus on Fabry disease. Wiley Interdisciplinary Reviews: Nanomedicine and Nanobiotechnology, 2021, 13, e1684.	6.1	12
34	Biomarkers in Fabry Disease. Implications for Clinical Diagnosis and Follow-up. Journal of Clinical Medicine, 2021, 10, 1664.	2.4	12
35	Long-term outcomes with agalsidase alfa enzyme replacement therapy: Analysis using deconstructed composite events. Molecular Genetics and Metabolism Reports, 2018, 14, 31-35.	1.1	11
36	Elosulfase alfa for mucopolysaccharidosis type IVA: Real-world experience in 7 patients from the Spanish Morquio-A early access program. Molecular Genetics and Metabolism Reports, 2018, 15, 116-120.	1.1	10

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37	A 15-Year Perspective of the Fabry Outcome Survey. FIRE Forum for International Research in Education, 2016, 4, 232640981666629.	0.7	9
38	Gender Differences in the Application of Spanish Criteria for Initiation of Enzyme Replacement Therapy for Fabry Disease in the Fabry Outcome Survey. International Journal of Molecular Sciences, 2016, 17, 1965.	4.1	7
39	Characterization of RAN Translation and Antisense Transcription in Primary Cell Cultures of Patients with Myotonic Dystrophy Type 1. Journal of Clinical Medicine, 2021, 10, 5520.	2.4	7
40	Cytochrome c Oxidase Deficiency in Muscle With Dicarboxylic Aciduria and Renal Tubular Acidosis. Journal of Child Neurology, 1990, 5, 147-152.	1.4	5
41	The Need for Establishing a Universal CTG Sizing Method in Myotonic Dystrophy Type 1. Genes, 2020, 11, 757.	2.4	5
42	Clinical features and health-related quality of life in adult patients with mucopolysaccharidosis IVA: the Spanish experience. Orphanet Journal of Rare Diseases, 2021, 16, 464.	2.7	5
43	Increased monocyte-dependent suppression of polyclonal activation of B lymphocytes from cystinotic children. Pediatric Nephrology, 1991, 5, 597-602.	1.7	4
44	Manifesting heterozygotes in McArdle disease: a myth or a realityâ€"role of statins. Journal of Inherited Metabolic Disease, 2018, 41, 1027-1035.	3.6	4
45	Preliminary Findings on CTG Expansion Determination in Different Tissues from Patients with Myotonic Dystrophy Type 1. Genes, 2020, 11, 1321.	2.4	4
46	Myotilinopathy unmasked by statin treatment: A case report. Muscle and Nerve, 2018, 57, E138-E140.	2.2	3
47	Quantification of urinary derivatives of Phenylbutyric and Benzoic acids by LC-MS/MS as treatment compliance biomarkers in Urea Cycle disorders. Journal of Pharmaceutical and Biomedical Analysis, 2019, 176, 112798.	2.8	3
48	Targeted nanoliposomes for the treatment of Fabry disease. Molecular Genetics and Metabolism, 2019, 126, S17.	1.1	3
49	Eating disorder in a patient with phenotypical features of Lujanâ€"Fryns syndrome. Clinical Dysmorphology, 2006, 15, 181-184.	0.3	1
50	Some questions about the transtubular potassium concentration gradient. Pediatric Nephrology, 1991, 5, 94-95.	1.7	0
51	Management of Fabry Disease with Agalsidase Treatment. Clinical Medicine Insights Therapeutics, 2010, 2, CMT.S6104.	0.4	0
52	Urinary levels of regenerating protein Î± do not differentiate celiac patients and healthy subjects. Biomarkers, 2013, 18, 178-180.	1.9	0