Maria Luz Couce

List of Publications by Citations

149 papers	2,611 citations	26 h-index	43 g-index
185 ext. papers	3,384 ext. citations	3.7 avg, IF	4.92 L-index

#	Paper	IF	Citations
149	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1041-57	5.4	143
148	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1059-74	5.4	135
147	Guidelines for diagnosis and management of the cobalamin-related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. *Journal of Inherited Metabolic Disease*, **2017**, 40, 21-48	5.4	126
146	A glimpse into past, present, and future DNA sequencing. *Molecular Genetics and Metabolism*, **2013**, 110, 3-24	3.7	106
145	Automated therapy preparation of isoleucine formulations using 3D printing for the treatment of MSUD: First single-centre, prospective, crossover study in patients. *International Journal of Pharmaceutics*, **2019**, 567, 118497	6.5	91
144	Cross-sectional study of 168 patients with hepatorenal tyrosinaemia and implications for clinical practice. *Orphanet Journal of Rare Diseases*, **2014**, 9, 107	4.2	83
143	NGS Technologies as a Turning Point in Rare Disease Research , Diagnosis and Treatment. *Current Medicinal Chemistry*, **2018**, 25, 404-432	4.3	67
142	Diversity of approaches to classic galactosemia around the world: a comparison of diagnosis, intervention, and outcomes. *Journal of Inherited Metabolic Disease*, **2012**, 35, 1037-49	5.4	67
141	Genetic and cellular studies of oxidative stress in methylmalonic aciduria (MMA) cobalamin deficiency type C (cblC) with homocystinuria (MMACHC). *Human Mutation*, **2009**, 30, 1558-66	4.7	67
140	Sanfilippo syndrome: Overall review. *Pediatrics International*, **2015**, 57, 331-8	1.2	57
139	Evaluation and long-term follow-up of infants with inborn errors of metabolism identified in an expanded screening programme. *Molecular Genetics and Metabolism*, **2011**, 104, 470-5	3.7	57
138	Long-term follow-up and outcome of phenylketonuria patients on sapropterin: a retrospective study. *Pediatrics*, **2013**, 131, e1881-8	7.4	54
137	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. *Journal of Inherited Metabolic Disease*, **2016**, 39, 661-672	5.4	42
136	Oral Administration to Nursing Women of Lactobacillus fermentum CECT5716 Prevents Lactational Mastitis Development: A Randomized Controlled Trial. *Breastfeeding Medicine*, **2017**, 12, 202-209	2.1	35
135	Assessment of a targeted resequencing assay as a support tool in the diagnosis of lysosomal storage disorders. *Orphanet Journal of Rare Diseases*, **2014**, 9, 59	4.2	35
134	Glutaric aciduria type I: outcome of patients with early- versus late-diagnosis. *European Journal of Paediatric Neurology*, **2013**, 17, 383-9	3.8	34
133	AZATAX: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2-CDG). *Annals of Neurology*, **2019**, 85, 740-751	9.4	32

132	Risk factors for developing mineral bone disease in phenylketonuric patients. *Molecular Genetics and Metabolism*, **2013**, 108, 149-54	3.7	31
131	A novel stop mutation in the vascular endothelial growth factor-C gene (VEGFC) results in Milroy-like disease. *Journal of Medical Genetics*, **2014**, 51, 475-8	5.8	31
130	Effects of different arachidonic acid supplementation on psychomotor development in very preterm infants; a randomized controlled trial. *Nutrition Journal*, **2015**, 14, 101	4.3	30
129	Tyrosinemia type 1 in Spain: mutational analysis, treatment and long-term outcome. *Pediatrics International*, **2011**, 53, 985-9	1.2	29
128	Carglumic acid enhances rapid ammonia detoxification in classical organic acidurias with a favourable risk-benefit profile: a retrospective observational study. *Orphanet Journal of Rare Diseases*, **2016**, 11, 32	4.2	29
127	Vitamin and mineral status in patients with hyperphenylalaninemia. *Molecular Genetics and Metabolism*, **2015**, 115, 145-50	3.7	28
126	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: Data from the E-HOD registry. *Journal of Inherited Metabolic Disease*, **2019**, 42, 333-352	5.4	28
125	Mucopolysaccharidosis IVA: Diagnosis, Treatment, and Management. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	27
124	Urea cycle disorders in Spain: an observational, cross-sectional and multicentric study of 104 cases. *Orphanet Journal of Rare Diseases*, **2014**, 9, 187	4.2	26
123	Clinical and metabolic findings in patients with methionine adenosyltransferase I/III deficiency detected by newborn screening. *Molecular Genetics and Metabolism*, **2013**, 110, 218-21	3.7	25
122	Clinical, genetic, and therapeutic diversity in 2 patients with severe mevalonate kinase deficiency. *Pediatrics*, **2012**, 129, e535-9	7.4	25
121	Newborn screening for homocystinurias: Recent recommendations versus current practice. *Journal of Inherited Metabolic Disease*, **2019**, 42, 128-139	5.4	24
120	Anthropometric characteristics and nutrition in a cohort of PAH-deficient patients. *Clinical Nutrition* , **2014**, 33, 702-17	5.9	23
119	Free-access copy-number variant detection tools for targeted next-generation sequencing data. *Mutation Research - Reviews in Mutation Research*, **2019**, 779, 114-125	7	23
118	Evolution of maple syrup urine disease in patients diagnosed by newborn screening versus late diagnosis. *European Journal of Paediatric Neurology*, **2015**, 19, 652-9	3.8	22
117	Consensus guideline for the diagnosis and management of mannose phosphate isomerase-congenital disorder of glycosylation. *Journal of Inherited Metabolic Disease*, **2020**, 43, 671-693	5.4	21
116	Newborn screening for Fabry disease in the north-west of Spain. *European Journal of Pediatrics*, **2017**, 176, 1075-1081	4.1	21
115	The identification of novel mutations in the biotinidase gene using denaturing high pressure liquid chromatography (dHPLC). *Molecular Genetics and Metabolism*, **2010**, 100, 42-5	3.7	21

114	Spectrum of CBS mutations in 16 homocystinuric patients from the Iberian Peninsula: high prevalence of T191M and absence of I278T or G307S. *Human Mutation*, **2003**, 22, 103	4.7	21
113	Recombinant Bile Salt-Stimulated Lipase in Preterm Infant Feeding: A Randomized Phase 3 Study. *PLoS ONE*, **2016**, 11, e0156071	3.7	21
112	Homozygous truncating mutation in prenatally expressed skeletal isoform of TTN gene results in arthrogryposis multiplex congenita and myopathy without cardiac involvement. *Neuromuscular Disorders*, **2017**, 27, 188-192	2.9	20
111	The p.T191M mutation of the CBS gene is highly prevalent among homocystinuric patients from Spain, Portugal and South America. *Journal of Human Genetics*, **2006**, 51, 305-313	4.3	20
110	Molecular epidemiology, genotype-phenotype correlation and BH4 responsiveness in Spanish patients with phenylketonuria. *Journal of Human Genetics*, **2016**, 61, 731-44	4.3	20
109	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders-A successful strategy for clinical research of rare diseases. *Journal of Inherited Metabolic Disease*, **2019**, 42, 93-106	5.4	20
108	Clinical and molecular diagnosis of non-phosphomannomutase 2 N-linked congenital disorders of glycosylation in Spain. *Clinical Genetics*, **2019**, 95, 615-626	4	20
107	Effects of Prebiotic and Probiotic Supplementation on Lactase Deficiency and Lactose Intolerance: A Systematic Review of Controlled Trials. *Nutrients*, **2020**, 12,	6.7	19
106	Development of electrospray ionization tandem mass spectrometry methods for the study of a high number of urine markers of inborn errors of metabolism. *Rapid Communications in Mass Spectrometry*, **2012**, 26, 2131-44	2.2	19
105	Lipid profile status and other related factors in patients with Hyperphenylalaninaemia. *Orphanet Journal of Rare Diseases*, **2016**, 11, 123	4.2	19
104	Relevance of urinary S100B protein levels as a short-term prognostic biomarker in asphyxiated infants treated with hypothermia. *Medicine (United States)*, **2017**, 96, e8453	1.8	18
103	Effects of Dairy Product Consumption on Height and Bone Mineral Content in Children: A Systematic Review of Controlled Trials. *Advances in Nutrition*, **2019**, 10, S88-S96	10	18
102	Rare Variants in 48 Genes Account for 42% of Cases of Epilepsy With or Without Neurodevelopmental Delay in 246 Pediatric Patients. *Frontiers in Neuroscience*, **2019**, 13, 1135	5.1	17
101	Molecular analysis of mucopolysaccharidosis IVA (Morquio A) in Spain. *Molecular Genetics and Metabolism*, **2012**, 106, 196-201	3.7	17
100	Molecular mechanisms of appetite and obesity: a role for brain AMPK. *Clinical Science*, **2016**, 130, 1697-709	6.3	17
99	A novel missense mutation in GRIN2A causes a nonepileptic neurodevelopmental disorder. *Movement Disorders*, **2018**, 33, 992-999	7	16
98	Tetrahydrobiopterin therapy vs phenylalanine-restricted diet: impact on growth in PKU. *Molecular Genetics and Metabolism*, **2013**, 109, 331-8	3.7	16
97	Molecular-genetic characterization and rescue of a TSFM mutation causing childhood-onset ataxia and nonobstructive cardiomyopathy. *European Journal of Human Genetics*, **2016**, 25, 153-156	5.3	16

96	Molecular epidemiology and genotype-phenotype correlation in phenylketonuria patients from South Spain. *Journal of Human Genetics*, **2013**, 58, 279-84	4.3	16
95	Non-alcoholic fatty liver in hereditary fructose intolerance. *Clinical Nutrition*, **2020**, 39, 455-459	5.9	15
94	Cost-Effectiveness Analysis of a National Newborn Screening Program for Biotinidase Deficiency. *Pediatrics*, **2015**, 136, e424-32	7.4	14
93	Clinical manifestations in female carriers of mucopolysaccharidosis type II: a Spanish cross-sectional study. *Orphanet Journal of Rare Diseases*, **2013**, 8, 92	4.2	14
92	Molecular epidemiology and BH4-responsiveness in patients with phenylalanine hydroxylase deficiency from Galicia region of Spain. *Gene*, **2013**, 521, 100-4	3.8	12
91	New evidence for assessing tetrahydrobiopterin (BH(4)) responsiveness. *Metabolism: Clinical and Experimental*, **2012**, 61, 1809-16	12.7	12
90	Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	12
89	Longitudinal volumetric and 2D assessment of cerebellar atrophy in a large cohort of children with phosphomannomutase deficiency (PMM2-CDG). *Journal of Inherited Metabolic Disease*, **2017**, 40, 709-713	5.4	11
88	Betaine anhydrous in homocystinuria: results from the RoCH registry. *Orphanet Journal of Rare Diseases*, **2019**, 14, 66	4.2	11
87	Clinical and genetic features of 13 Spanish patients with KCNQ2 mutations. *Journal of Human Genetics*, **2017**, 62, 185-189	4.3	11
86	Arterial stiffness assessment in patients with phenylketonuria. *Medicine (United States)*, **2017**, 96, e9322	1.8	11
85	Tension pneumocephalus induced by high-flow nasal cannula ventilation in a neonate. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2017**, 102, F173-F175	4.7	10
84	Genotype and phenotype characterization in a Spanish cohort with isovaleric acidemia. *Journal of Human Genetics*, **2017**, 62, 355-360	4.3	10
83	The Effect of Morbidity and Sex on Postnatal Growth of Very Preterm Infants: A Multicenter Cohort Study. *Neonatology*, **2019**, 115, 348-354	4	10
82	New insights in growth of phenylketonuric patients. *European Journal of Pediatrics*, **2015**, 174, 651-9	4.1	10
81	Newborn screening for medium-chain acyl-CoA dehydrogenase deficiency: regional experience and high incidence of carnitine deficiency. *Orphanet Journal of Rare Diseases*, **2013**, 8, 102	4.2	10
80	A selective screening program for the early detection of mucopolysaccharidosis: Results of the FIND project - a 2-year follow-up study. *Medicine (United States)*, **2017**, 96, e6887	1.8	10
79	Infantile-onset Pompe disease with neonatal debut: A case report and literature review. *Medicine (United States)*, **2017**, 96, e9186	1.8	10

78	Similarities between acylcarnitine profiles in large for gestational age newborns and obesity. *Scientific Reports*, **2017**, 7, 16267	4.9	10
77	Inborn errors of metabolism in a neonatology unit: impact and long-term results. *Pediatrics International*, **2011**, 53, 13-7	1.2	10
76	Preclinical screening for retinopathy of prematurity risk using IGF1 levels at 3 weeks post-partum. *PLoS ONE*, **2014**, 9, e88781	3.7	10
75	Congenital intestinal atresias with multiple episodes of sepsis: A case report and review of literature. *Medicine (United States)*, **2018**, 97, e10939	1.8	9
74	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	5.3	9
73	Carbohydrate status in patients with phenylketonuria. *Orphanet Journal of Rare Diseases*, **2018**, 13, 103	4.2	9
72	Early NT-proBNP levels as a screening tool for the detection of hemodynamically significant patent ductus arteriosus during the first week of life in very low birth weight infants. *Journal of Perinatology*, **2018**, 38, 881-888	3.1	9
71	Nonketotic hyperglycinemia: Functional assessment of missense variants in GLDC to understand phenotypes of the disease. *Human Mutation*, **2017**, 38, 678-691	4.7	8
70	Early cardiac abnormalities in obese children and their relationship with adiposity. *Nutrition*, **2018**, 46, 83-89	4.8	8
69	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	1.8	8
68	Genes and Variants Underlying Human Congenital Lactic Acidosis-From Genetics to Personalized Treatment. *Journal of Clinical Medicine*, **2019**, 8,	5.1	8
67	Prenatal alcohol exposure and its repercussion on newborns. *Journal of Neonatal-Perinatal Medicine* , **2014**, 7, 47-54	1.3	8
66	Bone Status in Patients with Phenylketonuria: A Systematic Review. *Nutrients*, **2020**, 12,	6.7	8
65	Acylcarnitine profile in neonatal hypoxic-ischemic encephalopathy: The value of butyrylcarnitine as a prognostic marker. *Medicine (United States)*, **2019**, 98, e15221	1.8	8
64	6R-tetrahydrobiopterin treated PKU patients below 4 years of age: Physical outcomes, nutrition and genotype. *Molecular Genetics and Metabolism*, **2015**, 115, 10-6	3.7	7
63	A quantitative assessment of the evolution of cerebellar syndrome in children with phosphomannomutase-deficiency (PMM2-CDG). *Orphanet Journal of Rare Diseases*, **2017**, 12, 155	4.2	7
62	Prospective and Retrospective Diagnosis of Barth Syndrome Aided by Next-Generation Sequencing. *American Journal of Clinical Pathology*, **2016**, 145, 507-13	1.9	7
61	Newborn Screening for Homocystinuria Revealed a High Frequency of MAT I/III Deficiency in Iberian Peninsula. *JIMD Reports*, **2015**, 20, 113-20	1.9	7

60	Arginine-guanidinoacetate-creatine pathway in preterm newborns: creatine biosynthesis in newborns. *Journal of Pediatric Endocrinology and Metabolism*, **2013**, 26, 53-60	1.6	7
59	Neonatal lethal hypophosphatasia: A case report and review of literature. *Medicine (United States)*, **2018**, 97, e13269	1.8	7
58	BMP8 and activated brown adipose tissue in human newborns. *Nature Communications*, **2021**, 12, 5274	17.4	7
57	Proteomic Analysis in Morquio A Cells Treated with Immobilized Enzymatic Replacement Therapy on Nanostructured Lipid Systems. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	6
56	Evaluation of carnitine deficit in very low birth weight preterm newborns small for their gestational age. *Journal of Maternal-Fetal and Neonatal Medicine*, **2016**, 29, 933-7	2	6
55	Long-term pharmacological management of phenylketonuria, including patients below the age of 4 years. *JIMD Reports*, **2012**, 2, 91-6	1.9	6
54	Effects of Nutritional Education Interventions on Metabolic Risk in Children and Adolescents: A Systematic Review of Controlled Trials. *Nutrients*, **2019**, 12,	6.7	6
53	Evolution of tyrosinemia type 1 disease in patients treated with nitisinone in Spain. *Medicine (United States)*, **2019**, 98, e17303	1.8	6
52	Retrospective study to identify risk factors for chronic kidney disease in children with congenital solitary functioning kidney detected by neonatal renal ultrasound screening. *Medicine (United States)*, **2018**, 97, e11819	1.8	6
51	Cohort study showed that growth rate increment has not been enough to prevent growth retardation of preterm infants and raised concerns about unbalanced growth. *Acta Paediatrica, International Journal of Paediatrics*, **2019**, 108, 1793-1800	3.1	5
50	Influence of phenylketonuria® diet on dimethylated arginines and methylation cycle. *Medicine (United States)*, **2017**, 96, e7392	1.8	5
49	Micronutrient in hyperphenylalaninemia. *Data in Brief*, **2015**, 4, 614-21	1.2	5
48	Nutritional practices in very low birth weight infants: a national survey. *Nutricion Hospitalaria*, **2017**, 34, 1067-1072	1	5
47	Potential protective role of endogenous glutamate-oxaloacetate transaminase against glutamate excitotoxicity in fetal hypoxic-ischaemic asphyxia. *Developmental Medicine and Child Neurology*, **2016**, 58, 57-62	3.3	5
46	Umbilical cord and visceral hemangiomas diagnosed in the neonatal period: A case report and a review of the literature. *Medicine (United States)*, **2016**, 95, e5196	1.8	5
45	Discovery of Biomarker Panels for Neural Dysfunction in Inborn Errors of Amino Acid Metabolism. *Scientific Reports*, **2019**, 9, 9128	4.9	4
44	Effects of LC-PUFA Supplementation in Patients with Phenylketonuria: A Systematic Review of Controlled Trials. *Nutrients*, **2019**, 11,	6.7	4
43	The early detection of Salla disease through second-tier tests in newborn screening: how to face incidental findings. *European Journal of Medical Genetics*, **2014**, 57, 527-31	2.6	4

42	Bone-Specific Drug Delivery for Osteoporosis and Rare Skeletal Disorders. *Current Osteoporosis Reports*, **2020**, 18, 515-525	5.4	4
41	Metabolic Bone Disease of Prematurity: Risk Factors and Associated Short-Term Outcomes. *Nutrients*, **2020**, 12,	6.7	4
40	Human Milk Concentrations of Minerals, Essential and Toxic Trace Elements and Association with Selective Medical, Social, Demographic and Environmental Factors. *Nutrients*, **2021**, 13,	6.7	4
39	Identification and Characterization of New Variants in Gene Expands the Clinical Spectrum Associated with Mitochondrial Complex I Deficiency. *Journal of Clinical Medicine*, **2019**, 8,	5.1	3
38	Enzyme-Loaded Gel Core Nanostructured Lipid Carriers to Improve Treatment of Lysosomal Storage Diseases: Formulation and In Vitro Cellular Studies of Elosulfase Alfa-Loaded Systems. *Pharmaceutics*, **2019**, 11,	6.4	3
37	Characterization of New Proteomic Biomarker Candidates in Mucopolysaccharidosis Type IVA. *International Journal of Molecular Sciences*, **2020**, 22,	6.3	3
36	Plasma Proteomic Analysis in Morquio A Disease. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
35	The Impact of Postnatal Systemic Steroids on the Growth of Preterm Infants: A Multicenter Cohort Study. *Nutrients*, **2019**, 11,	6.7	3
34	PattRec: An easy-to-use CNV detection tool optimized for targeted NGS assays with diagnostic purposes. *Genomics*, **2020**, 112, 1245-1256	4.3	3
33	Reply letter to "safety of SARS-Cov-2 vaccines administration for adult patients with hereditary fructose intolerance". *Human Vaccines and Immunotherapeutics*, **2021**, 1-2	4.4	3
32	Towards the automated economic assessment of newborn screening for rare diseases. *Journal of Biomedical Informatics*, **2019**, 95, 103216	10.2	2
31	Transition from paediatric care to adult care for patients with mucopolysaccharidosis. *Revista Clinica Espanola*, **2018**, 218, 17-21	0.7	2
30	Knee dislocation in the delivery room. *Journal of Pediatrics*, **2014**, 165, 871	3.6	2
29	A new case of maternal phenylketonuria treated with sapropterin dihydrochloride (6R-BH4). *Gynecological Endocrinology*, **2014**, 30, 691-3	2.4	2
28	Relevance of expanded neonatal screening of medium-chain acyl co-a dehydrogenase deficiency: outcome of a decade in galicia (Spain). *JIMD Reports*, **2011**, 1, 131-6	1.9	2
27	Glycogen Storage Disease Type Ia: Current Management Options, Burden and Unmet Needs. *Nutrients*, **2021**, 13,	6.7	2
26	Automated generation of decision-tree models for the economic assessment of interventions for rare diseases using the RaDiOS ontology. *Journal of Biomedical Informatics*, **2020**, 110, 103563	10.2	2
25	Rapid Phenotype-Driven Gene Sequencing with the NeoSeq Panel: A Diagnostic Tool for Critically Ill Newborns with Suspected Genetic Disease. *Journal of Clinical Medicine*, **2020**, 9,	5.1	2

24	Bone Mineral Density, Body Composition, and Metabolic Health of Very Low Birth Weight Infants Fed in Hospital Following Current Macronutrient Recommendations during the First 3 Years of Life. *Nutrients*, **2021**, 13,	6.7	2
23	Treatment adherence in tyrosinemia type 1 patients. *Orphanet Journal of Rare Diseases*, **2021**, 16, 256	4.2	2
22	Diagnosis and follow-up of patients with Hunter syndrome in Spain: A Delphi consensus. *Medicine (United States)*, **2018**, 97, e11246	1.8	2
21	Hepatic damage and glutamate oxaloacetate transaminase elevations during fetal asphyxia. *Developmental Medicine and Child Neurology*, **2017**, 59, 233-234	3.3	1
20	Home births: A growing phenomenon with potential risks. *Anales De Pediatr (English Edition)*, **2020**, 93, 266.e1-266.e6	0.4	1
19	Clinical Utility of Genotyping in Children with Suspected Functional Gastrointestinal Disorder. *Nutrients*, **2020**, 12,	6.7	1
18	Newborn screening for metabolic disorders in Spain and worldwide. *Anales De Pediatr (English Edition)*, **2019**, 91, 128e.1-128e.14	0.4	1
17	Progressive heterotopic ossification: the arduousness of an accurate diagnosis. *Journal of Pediatrics* , **2014**, 164, 203-4	3.6	1
16	Letter to the editor concerning the article Safety of vaccines administration in hereditary fructose intoleranceQ*Human Vaccines and Immunotherapeutics*, **2021**, 17, 2593-2594	4.4	1
15	Trace elements in dried blood spots as potential discriminating features for metabolic disorder diagnosis in newborns. *Metallomics*, **2021**, 13,	4.5	1
14	V232D mutation in patients with cystic fibrosis: Not so rare, not so mild. *Medicine (United States)*, **2018**, 97, e11397	1.8	1
13	Sedoanalgesia en las unidades neonatales. *Anales De Pediatr*, **2021**, 95, 126.e1-126.e11	0.2	1
12	The clinical and biochemical hallmarks generally associated with GLUT1DS may be caused by defects in genes other than SLC2A1.. *Clinical Genetics*, **2022**,	4	1
11	Fifty years of neonatal screening for congenital diseases in Spain. *Anales De Pediatr (English Edition)*, **2019**, 90, 205-206	0.4	0
10	Asymmetric dimethylarginine as a potential biomarker for management and follow-up of phenylketonuria. *European Journal of Pediatrics*, **2019**, 178, 903-911	4.1	0
9	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	4.2	0
8	Sedoanalgesia in neonatal units. *Anales De Pediatr (English Edition)*, **2021**, 95, 126.e1-126.e11	0.4	0
7	Rapid Molecular Diagnosis of Genetically Inherited Neuromuscular Disorders Using Next-Generation Sequencing Technologies. *Journal of Clinical Medicine*, **2022**, 11, 2750	5.1	0

6	Utility of bone turnover markers in metabolic bone disease detection in patients with phenylketonuria. *Medicina Clínica (English Edition)*, **2015**, 144, 193-197	0.3
5	Dimethylarginines as biomarkers for the kidney transplant management in methylmalonic aciduria. *Nephrology*, **2015**, 20, 576-9	2.2
4	Transocular Doppler ultrasonography of the central retinal artery is not an effective method for early diagnosis of retinopathy of prematurity. *Journal of Neonatal-Perinatal Medicine*, **2009**, 2, 163-167	1.3
3	Perinatal palliative care.. *Anales De Pediatría (English Edition)*, **2022**, 96, 60.e1-60.e1	0.4
2	Recommendations on the skills profile and standards of the neonatal transport system in Spain. *Anales De Pediatría (English Edition)*, **2021**, 94, 420.e1-420.e11	0.4
1	Transcriptomic analysis of patients with clinical suspicion of maturity-onset diabetes of the young (MODY) with a negative genetic diagnosis.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 105	4.2