

Maria Luz Couce

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Perinatal palliative care.. <i>Anales De Pediatr� (English Edition)</i> , 2022 , 96, 60.e1-60.e1	0.4	
148	Transcriptomic analysis of patients with clinical suspicion of maturity-onset diabetes of the young (MODY) with a negative genetic diagnosis.. <i>Orphanet Journal of Rare Diseases</i> , 2022 , 17, 105	4.2	
147	The clinical and biochemical hallmarks generally associated with GLUT1DS may be caused by defects in genes other than SLC2A1.. <i>Clinical Genetics</i> , 2022 ,	4	1
146	Rapid Molecular Diagnosis of Genetically Inherited Neuromuscular Disorders Using Next-Generation Sequencing Technologies. <i>Journal of Clinical Medicine</i> , 2022 , 11, 2750	5.1	0
145	Clinical features and health-related quality of life in adult patients with mucopolysaccharidosis IVA: the Spanish experience. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 464	4.2	0
144	Glycogen Storage Disease Type Ia: Current Management Options, Burden and Unmet Needs. <i>Nutrients</i> , 2021 , 13,	6.7	2
143	Letter to the editor concerning the article Safety of vaccines administration in hereditary fructose intoleranceQ <i>Human Vaccines and Immunotherapeutics</i> , 2021 , 17, 2593-2594	4.4	1
142	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. <i>Nutrients</i> , 2021 , 13,	6.7	2
141	Trace elements in dried blood spots as potential discriminating features for metabolic disorder diagnosis in newborns. <i>Metallomics</i> , 2021 , 13,	4.5	1
140	Human Milk Concentrations of Minerals, Essential and Toxic Trace Elements and Association with Selective Medical, Social, Demographic and Environmental Factors. <i>Nutrients</i> , 2021 , 13,	6.7	4
139	Recommendations on the skills profile and standards of the neonatal transport system in Spain. <i>Anales De Pediatr� (English Edition)</i> , 2021 , 94, 420.e1-420.e11	0.4	
138	Plasma Proteomic Analysis in Morquio A Disease. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	3
137	Treatment adherence in tyrosinemia type 1 patients. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 256	4.2	2
136	Sedoanalgesia in neonatal units. <i>Anales De Pediatr� (English Edition)</i> , 2021 , 95, 126.e1-126.e11	0.4	0
135	Sedoanalgesia en las unidades neonatales. <i>Anales De Pediatr�</i> , 2021 , 95, 126.e1-126.e11	0.2	1
134	Reply letter to "safety of SARS-Cov-2 vaccines administration for adult patients with hereditary fructose intolerance". <i>Human Vaccines and Immunotherapeutics</i> , 2021 , 1-2	4.4	3
133	BMP8 and activated brown adipose tissue in human newborns. <i>Nature Communications</i> , 2021 , 12, 5274	17.4	7

132	Home births: A growing phenomenon with potential risks. <i>Anales De Pediatr�� (English Edition)</i> , 2020 , 93, 266.e1-266.e6	0.4	1
131	Clinical Utility of Genotyping in Children with Suspected Functional Gastrointestinal Disorder. <i>Nutrients</i> , 2020 , 12,	6.7	1
130	Effects of Prebiotic and Probiotic Supplementation on Lactase Deficiency and Lactose Intolerance: A Systematic Review of Controlled Trials. <i>Nutrients</i> , 2020 , 12,	6.7	19
129	Mucopolysaccharidosis IVA: Diagnosis, Treatment, and Management. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	27
128	Consensus guideline for the diagnosis and management of mannose phosphate isomerase-congenital disorder of glycosylation. <i>Journal of Inherited Metabolic Disease</i> , 2020 , 43, 671-693	5.4	21
127	Characterization of New Proteomic Biomarker Candidates in Mucopolysaccharidosis Type IVA. <i>International Journal of Molecular Sciences</i> , 2020 , 22,	6.3	3
126	Automated generation of decision-tree models for the economic assessment of interventions for rare diseases using the RaDiOS ontology. <i>Journal of Biomedical Informatics</i> , 2020 , 110, 103563	10.2	2
125	Bone Status in Patients with Phenylketonuria: A Systematic Review. <i>Nutrients</i> , 2020 , 12,	6.7	8
124	Rapid Phenotype-Driven Gene Sequencing with the NeoSeq Panel: A Diagnostic Tool for Critically Ill Newborns with Suspected Genetic Disease. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	2
123	Bone-Specific Drug Delivery for Osteoporosis and Rare Skeletal Disorders. <i>Current Osteoporosis Reports</i> , 2020 , 18, 515-525	5.4	4
122	Metabolic Bone Disease of Prematurity: Risk Factors and Associated Short-Term Outcomes. <i>Nutrients</i> , 2020 , 12,	6.7	4
121	Non-alcoholic fatty liver in hereditary fructose intolerance. <i>Clinical Nutrition</i> , 2020 , 39, 455-459	5.9	15
120	PattRec: An easy-to-use CNV detection tool optimized for targeted NGS assays with diagnostic purposes. <i>Genomics</i> , 2020 , 112, 1245-1256	4.3	3
119	Identification and Characterization of New Variants in Gene Expands the Clinical Spectrum Associated with Mitochondrial Complex I Deficiency. <i>Journal of Clinical Medicine</i> , 2019 , 8,	5.1	3
118	Proteomic Analysis in Morquio A Cells Treated with Immobilized Enzymatic Replacement Therapy on Nanostructured Lipid Systems. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	6
117	Discovery of Biomarker Panels for Neural Dysfunction in Inborn Errors of Amino Acid Metabolism. <i>Scientific Reports</i> , 2019 , 9, 9128	4.9	4
116	Towards the automated economic assessment of newborn screening for rare diseases. <i>Journal of Biomedical Informatics</i> , 2019 , 95, 103216	10.2	2
115	Fifty years of neonatal screening for congenital diseases in Spain. <i>Anales De Pediatr�� (English Edition)</i> , 2019 , 90, 205-206	0.4	0

114	Cohort study showed that growth rate increment has not been enough to prevent growth retardation of preterm infants and raised concerns about unbalanced growth. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2019 , 108, 1793-1800	3.1	5
113	The Effect of Morbidity and Sex on Postnatal Growth of Very Preterm Infants: A Multicenter Cohort Study. <i>Neonatology</i> , 2019 , 115, 348-354	4	10
112	Betaine anhydrous in homocystinuria: results from the RoCH registry. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 66	4.2	11
111	AZATAx: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2-CDG). <i>Annals of Neurology</i> , 2019 , 85, 740-751	9.4	32
110	Asymmetric dimethylarginine as a potential biomarker for management and follow-up of phenylketonuria. <i>European Journal of Pediatrics</i> , 2019 , 178, 903-911	4.1	0
109	Newborn screening for homocystinurias: Recent recommendations versus current practice. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 128-139	5.4	24
108	Newborn screening for metabolic disorders in Spain and worldwide. <i>Anales De Pediatr�a (English Edition)</i> , 2019 , 91, 128e.1-128e.14	0.4	1
107	Effects of LC-PUFA Supplementation in Patients with Phenylketonuria: A Systematic Review of Controlled Trials. <i>Nutrients</i> , 2019 , 11,	6.7	4
106	Automated therapy preparation of isoleucine formulations using 3D printing for the treatment of MSUD: First single-centre, prospective, crossover study in patients. <i>International Journal of Pharmaceutics</i> , 2019 , 567, 118497	6.5	91
105	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. <i>Pharmaceutics</i> , 2019 , 11,	6.4	3
104	Rare Variants in 48 Genes Account for 42% of Cases of Epilepsy With or Without Neurodevelopmental Delay in 246 Pediatric Patients. <i>Frontiers in Neuroscience</i> , 2019 , 13, 1135	5.1	17
103	Genes and Variants Underlying Human Congenital Lactic Acidosis-From Genetics to Personalized Treatment. <i>Journal of Clinical Medicine</i> , 2019 , 8,	5.1	8
102	Effects of Dairy Product Consumption on Height and Bone Mineral Content in Children: A Systematic Review of Controlled Trials. <i>Advances in Nutrition</i> , 2019 , 10, S88-S96	10	18
101	Effects of Nutritional Education Interventions on Metabolic Risk in Children and Adolescents: A Systematic Review of Controlled Trials. <i>Nutrients</i> , 2019 , 12,	6.7	6
100	Free-access copy-number variant detection tools for targeted next-generation sequencing data. <i>Mutation Research - Reviews in Mutation Research</i> , 2019 , 779, 114-125	7	23
99	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: Data from the E-HOD registry. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 333-352	5.4	28
98	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders-A successful strategy for clinical research of rare diseases. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 93-106	5.4	20
97	The Impact of Postnatal Systemic Steroids on the Growth of Preterm Infants: A Multicenter Cohort Study. <i>Nutrients</i> , 2019 , 11,	6.7	3

96	Acylcarnitine profile in neonatal hypoxic-ischemic encephalopathy: The value of butyrylcarnitine as a prognostic marker. <i>Medicine (United States)</i> , 2019 , 98, e15221	1.8	8
95	Evolution of tyrosinemia type 1 disease in patients treated with nitisinone in Spain. <i>Medicine (United States)</i> , 2019 , 98, e17303	1.8	6
94	Clinical and molecular diagnosis of non-phosphomannomutase 2 N-linked congenital disorders of glycosylation in Spain. <i>Clinical Genetics</i> , 2019 , 95, 615-626	4	20
93	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	5.3	9
92	A novel missense mutation in GRIN2A causes a nonepileptic neurodevelopmental disorder. <i>Movement Disorders</i> , 2018 , 33, 992-999	7	16
91	Transition from paediatric care to adult care for patients with mucopolysaccharidosis. <i>Revista Clinica Espanola</i> , 2018 , 218, 17-21	0.7	2
90	Early cardiac abnormalities in obese children and their relationship with adiposity. <i>Nutrition</i> , 2018 , 46, 83-89	4.8	8
89	NGS Technologies as a Turning Point in Rare Disease Research , Diagnosis and Treatment. <i>Current Medicinal Chemistry</i> , 2018 , 25, 404-432	4.3	67
88	Congenital intestinal atresias with multiple episodes of sepsis: A case report and review of literature. <i>Medicine (United States)</i> , 2018 , 97, e10939	1.8	9
87	Elosulfase alfa for mucopolysaccharidosis type IVA: Real-world experience in 7 patients from the Spanish Morquio-A early access program. <i>Molecular Genetics and Metabolism Reports</i> , 2018 , 15, 116-120	1.8	8
86	Neonatal lethal hypophosphatasia: A case report and review of literature. <i>Medicine (United States)</i> , 2018 , 97, e13269	1.8	7
85	Retrospective study to identify risk factors for chronic kidney disease in children with congenital solitary functioning kidney detected by neonatal renal ultrasound screening. <i>Medicine (United States)</i> , 2018 , 97, e11819	1.8	6
84	Diagnosis and follow-up of patients with Hunter syndrome in Spain: A Delphi consensus. <i>Medicine (United States)</i> , 2018 , 97, e11246	1.8	2
83	V232D mutation in patients with cystic fibrosis: Not so rare, not so mild. <i>Medicine (United States)</i> , 2018 , 97, e11397	1.8	1
82	Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. <i>International Journal of Molecular Sciences</i> , 2018 , 19,	6.3	12
81	Carbohydrate status in patients with phenylketonuria. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 103	4.2	9
80	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. <i>Journal of Perinatology</i> , 2018 , 38, 881-888	3.1	9
79	Hepatic damage and glutamate oxaloacetate transaminase elevations during fetal asphyxia. <i>Developmental Medicine and Child Neurology</i> , 2017 , 59, 233-234	3.3	1

78	Tension pneumocephalus induced by high-flow nasal cannula ventilation in a neonate. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2017 , 102, F173-F175	4.7	10
77	Guidelines for diagnosis and management of the cobalamin-related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2017 , 40, 21-48	5.4	126
76	Genotype and phenotype characterization in a Spanish cohort with isovaleric acidemia. <i>Journal of Human Genetics</i> , 2017 , 62, 355-360	4.3	10
75	Longitudinal volumetric and 2D assessment of cerebellar atrophy in a large cohort of children with phosphomannomutase deficiency (PMM2-CDG). <i>Journal of Inherited Metabolic Disease</i> , 2017 , 40, 709-713	5.4	11
74	Oral Administration to Nursing Women of Lactobacillus fermentum CECT5716 Prevents Lactational Mastitis Development: A Randomized Controlled Trial. <i>Breastfeeding Medicine</i> , 2017 , 12, 202-209	2.1	35
73	Nonketotic hyperglycinemia: Functional assessment of missense variants in GLDC to understand phenotypes of the disease. <i>Human Mutation</i> , 2017 , 38, 678-691	4.7	8
72	A quantitative assessment of the evolution of cerebellar syndrome in children with phosphomannomutase-deficiency (PMM2-CDG). <i>Orphanet Journal of Rare Diseases</i> , 2017 , 12, 155	4.2	7
71	Influence of phenylketonuriaQ diet on dimethylated arginines and methylation cycle. <i>Medicine (United States)</i> , 2017 , 96, e7392	1.8	5
70	Relevance of urinary S100B protein levels as a short-term prognostic biomarker in asphyxiated infants treated with hypothermia. <i>Medicine (United States)</i> , 2017 , 96, e8453	1.8	18
69	Newborn screening for Fabry disease in the north-west of Spain. <i>European Journal of Pediatrics</i> , 2017 , 176, 1075-1081	4.1	21
68	A selective screening program for the early detection of mucopolysaccharidosis: Results of the FIND project - a 2-year follow-up study. <i>Medicine (United States)</i> , 2017 , 96, e6887	1.8	10
67	Homozygous truncating mutation in prenatally expressed skeletal isoform of TTN gene results in arthrogryposis multiplex congenita and myopathy without cardiac involvement. <i>Neuromuscular Disorders</i> , 2017 , 27, 188-192	2.9	20
66	Clinical and genetic features of 13 Spanish patients with KCNQ2 mutations. <i>Journal of Human Genetics</i> , 2017 , 62, 185-189	4.3	11
65	Arterial stiffness assessment in patients with phenylketonuria. <i>Medicine (United States)</i> , 2017 , 96, e9322	1.8	11
64	Infantile-onset Pompe disease with neonatal debut: A case report and literature review. <i>Medicine (United States)</i> , 2017 , 96, e9186	1.8	10
63	Similarities between acylcarnitine profiles in large for gestational age newborns and obesity. <i>Scientific Reports</i> , 2017 , 7, 16267	4.9	10
62	Nutritional practices in very low birth weight infants: a national survey. <i>Nutricion Hospitalaria</i> , 2017 , 34, 1067-1072	1	5
61	Prospective and Retrospective Diagnosis of Barth Syndrome Aided by Next-Generation Sequencing. <i>American Journal of Clinical Pathology</i> , 2016 , 145, 507-13	1.9	7

60	Evaluation of carnitine deficit in very low birth weight preterm newborns small for their gestational age. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2016 , 29, 933-7	2	6
59	Molecular-genetic characterization and rescue of a TSFM mutation causing childhood-onset ataxia and nonobstructive cardiomyopathy. <i>European Journal of Human Genetics</i> , 2016 , 25, 153-156	5.3	16
58	Lipid profile status and other related factors in patients with Hyperphenylalaninaemia. <i>Orphanet Journal of Rare Diseases</i> , 2016 , 11, 123	4.2	19
57	Recombinant Bile Salt-Stimulated Lipase in Preterm Infant Feeding: A Randomized Phase 3 Study. <i>PLoS ONE</i> , 2016 , 11, e0156071	3.7	21
56	Potential protective role of endogenous glutamate-oxaloacetate transaminase against glutamate excitotoxicity in fetal hypoxic-ischaemic asphyxia. <i>Developmental Medicine and Child Neurology</i> , 2016 , 58, 57-62	3.3	5
55	Umbilical cord and visceral hemangiomas diagnosed in the neonatal period: A case report and a review of the literature. <i>Medicine (United States)</i> , 2016 , 95, e5196	1.8	5
54	Carglumic acid enhances rapid ammonia detoxification in classical organic acidurias with a favourable risk-benefit profile: a retrospective observational study. <i>Orphanet Journal of Rare Diseases</i> , 2016 , 11, 32	4.2	29
53	Molecular epidemiology, genotype-phenotype correlation and BH4 responsiveness in Spanish patients with phenylketonuria. <i>Journal of Human Genetics</i> , 2016 , 61, 731-44	4.3	20
52	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2016 , 39, 661-672	5.4	42
51	Molecular mechanisms of appetite and obesity: a role for brain AMPK. <i>Clinical Science</i> , 2016 , 130, 1697-709	3.9	17
50	Cost-Effectiveness Analysis of a National Newborn Screening Program for Biotinidase Deficiency. <i>Pediatrics</i> , 2015 , 136, e424-32	7.4	14
49	Sanfilippo syndrome: Overall review. <i>Pediatrics International</i> , 2015 , 57, 331-8	1.2	57
48	New insights in growth of phenylketonuric patients. <i>European Journal of Pediatrics</i> , 2015 , 174, 651-9	4.1	10
47	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. <i>Journal of Inherited Metabolic Disease</i> , 2015 , 38, 1041-57	5.4	143
46	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. <i>Journal of Inherited Metabolic Disease</i> , 2015 , 38, 1059-74	5.4	135
45	6R-tetrahydrobiopterin treated PKU patients below 4 years of age: Physical outcomes, nutrition and genotype. <i>Molecular Genetics and Metabolism</i> , 2015 , 115, 10-6	3.7	7
44	Evolution of maple syrup urine disease in patients diagnosed by newborn screening versus late diagnosis. <i>European Journal of Paediatric Neurology</i> , 2015 , 19, 652-9	3.8	22
43	Vitamin and mineral status in patients with hyperphenylalaninemia. <i>Molecular Genetics and Metabolism</i> , 2015 , 115, 145-50	3.7	28

42	Utility of bone turnover markers in metabolic bone disease detection in patients with phenylketonuria. <i>Medicina Clínica (English Edition)</i> , 2015 , 144, 193-197	0.3	
41	Micronutrient in hyperphenylalaninemia. <i>Data in Brief</i> , 2015 , 4, 614-21	1.2	5
40	Effects of different arachidonic acid supplementation on psychomotor development in very preterm infants; a randomized controlled trial. <i>Nutrition Journal</i> , 2015 , 14, 101	4.3	30
39	Dimethylarginines as biomarkers for the kidney transplant management in methylmalonic aciduria. <i>Nephrology</i> , 2015 , 20, 576-9	2.2	
38	Newborn Screening for Homocystinuria Revealed a High Frequency of MAT I/III Deficiency in Iberian Peninsula. <i>JIMD Reports</i> , 2015 , 20, 113-20	1.9	7
37	Anthropometric characteristics and nutrition in a cohort of PAH-deficient patients. <i>Clinical Nutrition</i> , 2014 , 33, 702-17	5.9	23
36	Cross-sectional study of 168 patients with hepatorenal tyrosinaemia and implications for clinical practice. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 107	4.2	83
35	Knee dislocation in the delivery room. <i>Journal of Pediatrics</i> , 2014 , 165, 871	3.6	2
34	Assessment of a targeted resequencing assay as a support tool in the diagnosis of lysosomal storage disorders. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 59	4.2	35
33	The early detection of Salla disease through second-tier tests in newborn screening: how to face incidental findings. <i>European Journal of Medical Genetics</i> , 2014 , 57, 527-31	2.6	4
32	Urea cycle disorders in Spain: an observational, cross-sectional and multicentric study of 104 cases. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 187	4.2	26
31	A new case of maternal phenylketonuria treated with sapropterin dihydrochloride (6R-BH4). <i>Gynecological Endocrinology</i> , 2014 , 30, 691-3	2.4	2
30	A novel stop mutation in the vascular endothelial growth factor-C gene (VEGFC) results in Milroy-like disease. <i>Journal of Medical Genetics</i> , 2014 , 51, 475-8	5.8	31
29	Prenatal alcohol exposure and its repercussion on newborns. <i>Journal of Neonatal-Perinatal Medicine</i> , 2014 , 7, 47-54	1.3	8
28	Progressive heterotopic ossification: the arduousness of an accurate diagnosis. <i>Journal of Pediatrics</i> , 2014 , 164, 203-4	3.6	1
27	Preclinical screening for retinopathy of prematurity risk using IGF1 levels at 3 weeks post-partum. <i>PLoS ONE</i> , 2014 , 9, e88781	3.7	10
26	Newborn screening for medium-chain acyl-CoA dehydrogenase deficiency: regional experience and high incidence of carnitine deficiency. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 102	4.2	10
25	Clinical manifestations in female carriers of mucopolysaccharidosis type II: a Spanish cross-sectional study. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 92	4.2	14

24	Tetrahydrobiopterin therapy vs phenylalanine-restricted diet: impact on growth in PKU. <i>Molecular Genetics and Metabolism</i> , 2013 , 109, 331-8	3.7	16
23	A glimpse into past, present, and future DNA sequencing. <i>Molecular Genetics and Metabolism</i> , 2013 , 110, 3-24	3.7	106
22	Long-term follow-up and outcome of phenylketonuria patients on sapropterin: a retrospective study. <i>Pediatrics</i> , 2013 , 131, e1881-8	7.4	54
21	Glutaric aciduria type I: outcome of patients with early- versus late-diagnosis. <i>European Journal of Paediatric Neurology</i> , 2013 , 17, 383-9	3.8	34
20	Clinical and metabolic findings in patients with methionine adenosyltransferase I/III deficiency detected by newborn screening. <i>Molecular Genetics and Metabolism</i> , 2013 , 110, 218-21	3.7	25
19	Risk factors for developing mineral bone disease in phenylketonuric patients. <i>Molecular Genetics and Metabolism</i> , 2013 , 108, 149-54	3.7	31
18	Molecular epidemiology and BH4-responsiveness in patients with phenylalanine hydroxylase deficiency from Galicia region of Spain. <i>Gene</i> , 2013 , 521, 100-4	3.8	12
17	Molecular epidemiology and genotype-phenotype correlation in phenylketonuria patients from South Spain. <i>Journal of Human Genetics</i> , 2013 , 58, 279-84	4.3	16
16	Arginine-guanidinoacetate-creatine pathway in preterm newborns: creatine biosynthesis in newborns. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2013 , 26, 53-60	1.6	7
15	Diversity of approaches to classic galactosemia around the world: a comparison of diagnosis, intervention, and outcomes. <i>Journal of Inherited Metabolic Disease</i> , 2012 , 35, 1037-49	5.4	67
14	New evidence for assessing tetrahydrobiopterin (BH(4)) responsiveness. <i>Metabolism: Clinical and Experimental</i> , 2012 , 61, 1809-16	12.7	12
13	Molecular analysis of mucopolysaccharidosis IVA (Morquio A) in Spain. <i>Molecular Genetics and Metabolism</i> , 2012 , 106, 196-201	3.7	17
12	Development of electrospray ionization tandem mass spectrometry methods for the study of a high number of urine markers of inborn errors of metabolism. <i>Rapid Communications in Mass Spectrometry</i> , 2012 , 26, 2131-44	2.2	19
11	Long-term pharmacological management of phenylketonuria, including patients below the age of 4 years. <i>JIMD Reports</i> , 2012 , 2, 91-6	1.9	6
10	Clinical, genetic, and therapeutic diversity in 2 patients with severe mevalonate kinase deficiency. <i>Pediatrics</i> , 2012 , 129, e535-9	7.4	25
9	Evaluation and long-term follow-up of infants with inborn errors of metabolism identified in an expanded screening programme. <i>Molecular Genetics and Metabolism</i> , 2011 , 104, 470-5	3.7	57
8	Inborn errors of metabolism in a neonatology unit: impact and long-term results. <i>Pediatrics International</i> , 2011 , 53, 13-7	1.2	10
7	Tyrosinemia type 1 in Spain: mutational analysis, treatment and long-term outcome. <i>Pediatrics International</i> , 2011 , 53, 985-9	1.2	29

6	Relevance of expanded neonatal screening of medium-chain acyl co-a dehydrogenase deficiency: outcome of a decade in galicia (Spain). <i>JIMD Reports</i> , 2011 , 1, 131-6	1.9	2
5	The identification of novel mutations in the biotinidase gene using denaturing high pressure liquid chromatography (dHPLC). <i>Molecular Genetics and Metabolism</i> , 2010 , 100, 42-5	3.7	21
4	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, 1558-66	4.7	67
3	Transocular Doppler ultrasonography of the central retinal artery is not an effective method for early diagnosis of retinopathy of prematurity. <i>Journal of Neonatal-Perinatal Medicine</i> , 2009 , 2, 163-167	1.3	
2	The p.T191M mutation of the CBS gene is highly prevalent among homocystinuric patients from Spain, Portugal and South America. <i>Journal of Human Genetics</i> , 2006 , 51, 305-313	4.3	20
1	Spectrum of CBS mutations in 16 homocystinuric patients from the Iberian Peninsula: high prevalence of T191M and absence of I278T or G307S. <i>Human Mutation</i> , 2003 , 22, 103	4.7	21