

# Giancarlo La Marca

## List of Publications by Year in descending order

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Version: 2024-02-01

136  
papers

5,373  
citations

61857

43  
h-index

106150

65  
g-index

139  
all docs

139  
docs citations

139  
times ranked

7108  
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. <i>Genetics in Medicine</i> , 2011, 13, 230-254.	1.1	308
2	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.	1.7	206
3	The "headache tree"™ via umbellulone and TRPA1 activates the trigeminovascular system. <i>Brain</i> , 2012, 135, 376-390.	3.7	163
4	The pathophysiology of retinopathy of prematurity: an update of previous and recent knowledge. <i>Acta Ophthalmologica</i> , 2014, 92, 2-20.	0.6	146
5	Role of the Adrenergic System in a Mouse Model of Oxygen-Induced Retinopathy: Antiangiogenic Effects of Î²-Adrenoreceptor Blockade. , 2011, 52, 155.		141
6	Odorant-Binding Proteins and Chemosensory Proteins in Pheromone Detection and Release in the Silkworm <i>Bombyx mori</i> . <i>Chemical Senses</i> , 2011, 36, 335-344.	1.1	134
7	Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. <i>International Journal of Neonatal Screening</i> , 2021, 7, 15.	1.2	118
8	Enhanced interpretation of newborn screening results without analyte cutoff values. <i>Genetics in Medicine</i> , 2012, 14, 648-655.	1.1	117
9	Proteinuria Impairs Podocyte Regeneration by Sequestering Retinoic Acid. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 1756-1768.	3.0	116
10	Expanded newborn screening by mass spectrometry: New tests, future perspectives. <i>Mass Spectrometry Reviews</i> , 2016, 35, 71-84.	2.8	107
11	Rapid 2nd-Tier Test for Measurement of 3-OH-Propionic and Methylmalonic Acids on Dried Blood Spots: Reducing the False-Positive Rate for Propionylcarnitine during Expanded Newborn Screening by Liquid Chromatography-Tandem Mass Spectrometry. <i>Clinical Chemistry</i> , 2007, 53, 1364-1369.	1.5	104
12	Oleuropein aglycone protects against pyroglutamylation-3 amyloid-Å toxicity: biochemical, epigenetic and functional correlates. <i>Neurobiology of Aging</i> , 2015, 36, 648-663.	1.5	91
13	Rapid assay of topiramate in dried blood spots by a new liquid chromatography-tandem mass spectrometric method. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2008, 48, 1392-1396.	1.4	87
14	Conventional and long-circulating liposomes of artemisinin: preparation, characterization, and pharmacokinetic profile in mice. <i>Journal of Liposome Research</i> , 2011, 21, 237-244.	1.5	87
15	Oral Propranolol for Retinopathy of Prematurity: Risks, Safety Concerns, and Perspectives. <i>Journal of Pediatrics</i> , 2013, 163, 1570-1577.e6.	0.9	80
16	Phenobarbital for neonatal seizures in hypoxic ischemic encephalopathy: A pharmacokinetic study during whole body hypothermia. <i>Epilepsia</i> , 2011, 52, 794-801.	2.6	79
17	Oleuropein, the Main Polyphenol of <i>Olea europaea</i> Leaf Extract, Has an Anti-Cancer Effect on Human BRAF Melanoma Cells and Potentiates the Cytotoxicity of Current Chemotherapies. <i>Nutrients</i> , 2018, 10, 1950.	1.7	79
18	Dominulin A and B: Two new antibacterial peptides identified on the cuticle and in the venom of the social paper wasp <i>Polistes dominulus</i> using MALDI-TOF, MALDI-TOF/TOF, and ESI-ion trap. <i>Journal of the American Society for Mass Spectrometry</i> , 2006, 17, 376-383.	1.2	78

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19	Topiramate concentrations in neonates treated with prolonged whole body hypothermia for hypoxic ischemic encephalopathy. <i>Epilepsia</i> , 2009, 50, 2355-2361.	2.6	75
20	A Chaperone Enhances Blood $\alpha$ -Glucosidase Activity in Pompe Disease Patients Treated With Enzyme Replacement Therapy. <i>Molecular Therapy</i> , 2014, 22, 2004-2012.	3.7	75
21	Hematopoietic Stem- and Progenitor-Cell Gene Therapy for Hurler Syndrome. <i>New England Journal of Medicine</i> , 2021, 385, 1929-1940.	13.9	75
22	Serum Levels of Acyl-Carnitines along the Continuum from Normal to Alzheimer's Dementia. <i>PLoS ONE</i> , 2016, 11, e0155694.	1.1	72
23	New Strategy for the Screening of Lysosomal Storage Disorders: The Use of the Online Trapping-and-Cleanup Liquid Chromatography/Mass Spectrometry. <i>Analytical Chemistry</i> , 2009, 81, 6113-6121.	3.2	65
24	Tandem mass spectrometry, but not T-cell receptor excision circle analysis, identifies newborns with late-onset adenosine deaminase deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1604-1610.	1.5	65
25	Oral Topiramate in Neonates with Hypoxic Ischemic Encephalopathy Treated with Hypothermia: A Safety Study. <i>Journal of Pediatrics</i> , 2010, 157, 361-366.	0.9	64
26	Are Patients with Potential Celiac Disease Really Potential? The Answer of Metabonomics. <i>Journal of Proteome Research</i> , 2011, 10, 714-721.	1.8	64
27	Neonatal screening for severe combined immunodeficiency caused by an adenosine deaminase defect: A reliable and inexpensive method using tandem mass spectrometry. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 127, 1394-1399.	1.5	63
28	$\alpha$ -Carbamylglutamate in Emergency Management of Hyperammonemia in Neonatal Acute Onset Propionic and Methylmalonic Aciduria. <i>Neonatology</i> , 2010, 97, 286-290.	0.9	60
29	A TRPA1 antagonist reverts oxaliplatin-induced neuropathic pain. <i>Scientific Reports</i> , 2013, 3, 2005.	1.6	58
30	The inclusion of succinylacetone as marker for tyrosinemia type I in expanded newborn screening programs. <i>Rapid Communications in Mass Spectrometry</i> , 2008, 22, 812-818.	0.7	57
31	Diagnosis of immunodeficiency caused by a purine nucleoside phosphorylase defect by using tandem mass spectrometry on dried blood spots. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 155-159.e3.	1.5	56
32	Mass spectrometry in clinical chemistry: the case of newborn screening. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2014, 101, 174-182.	1.4	55
33	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 273-283.	1.7	55
34	New developments in the treatment of hyperammonemia: emerging use of carglumic acid. <i>International Journal of General Medicine</i> , 2011, 4, 21.	0.8	54
35	Lactate Rewires Lipid Metabolism and Sustains a Metabolic Epigenetic Axis in Prostate Cancer. <i>Cancer Research</i> , 2022, 82, 1267-1282.	0.4	52
36	Rapid quantitation of globotriaosylceramide in human plasma and urine: a potential application for monitoring enzyme replacement therapy in Anderson-Fabry disease. <i>Rapid Communications in Mass Spectrometry</i> , 2002, 16, 1507-1514.	0.7	51

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37	A new rapid micromethod for the assay of phenobarbital from dried blood spots by LC-MS/MS. <i>Epilepsia</i> , 2009, 50, 2658-2662.	2.6	50
38	Study protocol: safety and efficacy of propranolol in newborns with Retinopathy of Prematurity (PROP-ROP): ISRCTN18523491. <i>BMC Pediatrics</i> , 2010, 10, 83.	0.7	50
39	Serum Amino Acid Profiles in Normal Subjects and in Patients with or at Risk of Alzheimer Dementia. <i>Dementia and Geriatric Cognitive Disorders Extra</i> , 2017, 7, 143-159.	0.6	50
40	Safety and efficacy of topiramate in neonates with hypoxic ischemic encephalopathy treated with hypothermia (NeoNATI): a feasibility study. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2018, 31, 973-980.	0.7	50
41	Development of an UPLC-MS/MS method for the determination of antibiotic ertapenem on dried blood spots. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2012, 61, 108-113.	1.4	49
42	Reticular dysgenesis-associated AK2 protects hematopoietic stem and progenitor cell development from oxidative stress. <i>Journal of Experimental Medicine</i> , 2015, 212, 1185-1202.	4.2	49
43	Adrenoceptor as a potential immunosuppressor agent in melanoma. <i>British Journal of Pharmacology</i> , 2019, 176, 2509-2524.	2.7	49
44	Rapid assay of rufinamide in dried blood spots by a new liquid chromatography-MS/MS method. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2011, 54, 192-197.	1.4	48
45	Delayed-onset adenosine deaminase deficiency: Strategies for an early diagnosis. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 130, 991-994.	1.5	44
46	Hypocitrullinemia in expanded newborn screening by LC-MS/MS is not a reliable marker for ornithine transcarbamylase deficiency. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2009, 49, 1292-1295.	1.4	43
47	Eye drop propranolol administration promotes the recovery of oxygen-induced retinopathy in mice. <i>Experimental Eye Research</i> , 2013, 111, 27-35.	1.2	43
48	Detection of honeybee venom in envenomed tissues by direct MALDI MSI. <i>Journal of the American Society for Mass Spectrometry</i> , 2009, 20, 112-123.	1.2	42
49	Rapid and sensitive LC-MS/MS method for the analysis of antibiotic linezolid on dried blood spot. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2012, 67-68, 86-91.	1.4	41
50	Safety and Tolerability of Antiepileptic Drug Treatment in Children with Epilepsy. <i>Drug Safety</i> , 2012, 35, 519-533.	1.4	41
51	Synergy between the pharmacological chaperone 1-deoxygalactonojirimycin and the human recombinant alpha-galactosidase A in cultured fibroblasts from patients with Fabry disease. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 513-520.	1.7	40
52	The inclusion of ADA-SCID in expanded newborn screening by tandem mass spectrometry. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2014, 88, 201-206.	1.4	40
53	Ultrasensitive detection of cancer biomarkers by nickel-based isolation of polydisperse extracellular vesicles from blood. <i>EBioMedicine</i> , 2019, 43, 114-126.	2.7	40
54	Therapeutic drug monitoring of carbamazepine and its metabolite in children from dried blood spots using liquid chromatography and tandem mass spectrometry. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2015, 109, 164-170.	1.4	39

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55	LC-MS/MS Method for Simultaneous Determination on a Dried Blood Spot of Multiple Analytes Relevant for Treatment Monitoring in Patients with Tyrosinemia Type I. <i>Analytical Chemistry</i> , 2012, 84, 1184-1188.	3.2	37
56	Newborn screening for homocystinurias: Recent recommendations versus current practice. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 128-139.	1.7	37
57	Implementing tandem mass spectrometry as a routine tool for characterizing the complete purine and pyrimidine metabolic profile in urine samples. <i>Journal of Mass Spectrometry</i> , 2006, 41, 1442-1452.	0.7	36
58	Solid Olive Residues: An Insight into Their Phenolic Composition. <i>Journal of Agricultural and Food Chemistry</i> , 2005, 53, 8963-8969.	2.4	34
59	Propranolol concentrations after oral administration in term and preterm neonates. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2013, 26, 833-840.	0.7	34
60	Pre- and post-dialysis quantitative dosage of thymidine in urine and plasma of a MNGIE patient by using HPLC-ESI-MS/MS. <i>Journal of Mass Spectrometry</i> , 2006, 41, 586-592.	0.7	33
61	Propranolol 0.2% Eye Micro-Drops for Retinopathy of Prematurity: A Prospective Phase IIB Study. <i>Frontiers in Pediatrics</i> , 2019, 7, 180.	0.9	31
62	Development of Strategies to Decrease False Positive Results in Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2020, 6, 84.	1.2	31
63	High-performance liquid chromatography/electrospray ionization tandem mass spectrometric investigation of stilbenoids in cell cultures of <i>Vitis vinifera</i> L., cv. Malvasia. <i>Rapid Communications in Mass Spectrometry</i> , 2010, 24, 2065-2073.	0.7	29
64	Food supplements of <i>Tribulus terrestris</i> L.: An HPLC-ESI-MS method for an estimation of the saponin content. <i>Chromatographia</i> , 2003, 57, 581-592.	0.7	28
65	Falsely elevated C4-carnitine as expression of glutamate formiminotransferase deficiency in tandem mass spectrometry newborn screening. <i>Journal of Mass Spectrometry</i> , 2006, 41, 263-265.	0.7	28
66	Safety and efficacy of topiramate in neonates with hypoxic ischemic encephalopathy treated with hypothermia (NeoNATI). <i>BMC Pediatrics</i> , 2012, 12, 144.	0.7	28
67	Fabry disease: polymorphic haplotypes and a novel missense mutation in the <i>GLA</i> gene. <i>Clinical Genetics</i> , 2012, 81, 224-233.	1.0	28
68	Sudden unexpected fatal encephalopathy in adults with OTC gene mutations-Clues for early diagnosis and timely treatment. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 105.	1.2	28
69	Heptadecanoylcarnitine (C17) a novel candidate biomarker for newborn screening of propionic and methylmalonic acidemias. <i>Clinica Chimica Acta</i> , 2015, 450, 342-348.	0.5	27
70	Clinical relevance of short-chain acyl-CoA dehydrogenase (SCAD) deficiency: Exploring the role of new variants including the first SCAD-disease-causing allele carrying a synonymous mutation. <i>BBA Clinical</i> , 2016, 5, 114-119.	4.1	27
71	Children who develop type 1 diabetes early in life show low levels of carnitine and amino acids at birth: does this finding shed light on the etiopathogenesis of the disease?. <i>Nutrition and Diabetes</i> , 2013, 3, e94-e94.	1.5	26
72	Electrospray ionisation tandem mass spectrometric investigation of phenylpropanoids and secoiridoids from solid olive residue. <i>Rapid Communications in Mass Spectrometry</i> , 2006, 20, 2013-2022.	0.7	25

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73	A rapid liquid chromatography tandem mass spectrometry-based method for measuring propranolol on dried blood spots. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2013, 78-79, 34-38.	1.4	25
74	Propranolol 0.1% eye micro-drops in newborns with retinopathy of prematurity: a pilot clinical trial. <i>Pediatric Research</i> , 2017, 81, 307-314.	1.1	24
75	Barth syndrome presenting with acute metabolic decompensation in the neonatal period. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 684-684.	1.7	22
76	Delivery of doxorubicin across the blood-brain barrier by ondansetron pretreatment: a study in vitro and in vivo. <i>Cancer Letters</i> , 2014, 353, 242-247.	3.2	22
77	Dried blood spot assay for the quantification of phenytoin using Liquid Chromatography-Mass Spectrometry. <i>Clinica Chimica Acta</i> , 2015, 440, 31-35.	0.5	22
78	Fatal Malonyl CoA Decarboxylase Deficiency Due to Maternal Uniparental Isodisomy of the Telomeric End of Chromosome 16. <i>Annals of Human Genetics</i> , 2007, 71, 705-712.	0.3	21
79	Detection of doxorubicin hydrochloride accumulation in the rat brain after morphine treatment by mass spectrometry. <i>Cancer Chemotherapy and Pharmacology</i> , 2011, 67, 1333-1340.	1.1	20
80	Rapid determination of orotic acid in urine by a fast liquid chromatography/tandem mass spectrometric method. <i>Rapid Communications in Mass Spectrometry</i> , 2003, 17, 788-793.	0.7	19
81	Simultaneous determination of creatine and guanidinoacetate in plasma by liquid chromatography-tandem mass spectrometry (LC-MS/MS). <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2011, 56, 792-798.	1.4	18
82	Strategies for reducing the incidence of skin complications in newborns treated with whole-body hypothermia. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2012, 25, 2115-2121.	0.7	18
83	Aminoacylase I deficiency due to <i>ACY1</i> mRNA exon skipping. <i>Clinical Genetics</i> , 2014, 86, 367-372.	1.0	18
84	Lipoic-Based TRPA1/TRPV1 Antagonist to Treat Orofacial Pain. <i>ACS Chemical Neuroscience</i> , 2015, 6, 380-385.	1.7	18
85	LC-MS/MS method for simultaneous quantification of heparan sulfate and dermatan sulfate in urine by butanolysis derivatization. <i>Clinica Chimica Acta</i> , 2019, 488, 98-103.	0.5	18
86	Estimating the integrity of aged DNA samples by CE. <i>Electrophoresis</i> , 2009, 30, 3986-3995.	1.3	17
87	Leukocyte and Dried Blood Spot Arylsulfatase A Assay by Tandem Mass Spectrometry. <i>Analytical Chemistry</i> , 2020, 92, 6341-6348.	3.2	17
88	Pharmacological modulation of blood-brain barrier increases permeability of doxorubicin into the rat brain. <i>American Journal of Cancer Research</i> , 2013, 3, 424-32.	1.4	17
89	Successful Propranolol Treatment of a Kaposiform Hemangioendothelioma Apparently Resistant to Propranolol. <i>Pediatric Blood and Cancer</i> , 2016, 63, 1290-1292.	0.8	16
90	The successful inclusion of succinylacetone as a marker of tyrosinemia type I in Tuscany newborn screening program. <i>Rapid Communications in Mass Spectrometry</i> , 2009, 23, 3891-3893.	0.7	15

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91	Measurement of succinyl-carnitine and methylmalonyl-carnitine on dried blood spot by liquid chromatography-tandem mass spectrometry. <i>Clinica Chimica Acta</i> , 2014, 429, 30-33.	0.5	15
92	Vacuolated PAS-positive lymphocytes as an hallmark of Pompe disease and other myopathies related to impaired autophagy. <i>Journal of Cellular Physiology</i> , 2018, 233, 5829-5837.	2.0	15
93	Pharmacokinetics and local safety profile of propranolol eye drops in rabbits. <i>Pediatric Research</i> , 2014, 76, 378-385.	1.1	14
94	Rapid diagnosis of medium chain Acyl Co-A dehydrogenase(MCAD) deficiency in a newborn by liquid chromatography/tandem mass spectrometry. <i>Rapid Communications in Mass Spectrometry</i> , 2003, 17, 2688-2692.	0.7	13
95	Thermal inactivation of SARS COVID-2 virus: Are steam inhalations a potential treatment?. <i>Life Sciences</i> , 2021, 265, 118801.	2.0	12
96	Analysis of Organic Acids and Acylglycines for the Diagnosis of Related Inborn Errors of Metabolism by GC- and HPLC-MS. <i>Methods in Molecular Biology</i> , 2011, 708, 73-98.	0.4	12
97	Synthesis of the essential core of the human glycosylphosphatidylinositol (GPI) anchor. <i>Bioorganic Chemistry</i> , 2011, 39, 88-93.	2.0	11
98	Newborn Screening for Tyrosinemia Type I: Further Evidence that Succinylacetone Determination on Blood Spot Is Essential. <i>JIMD Reports</i> , 2011, 1, 107-109.	0.7	11
99	Reference intervals for orotic acid in urine, plasma and dried blood spot using hydrophilic interaction liquid chromatography-tandem mass spectrometry. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2012, 883-884, 155-160.	1.2	11
100	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. <i>Brain</i> , 2022, 145, 2687-2703.	3.7	11
101	High frequency of biotinidase deficiency in Italian population identified by newborn screening. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100689.	0.4	10
102	Hyperhydroxyprolinaemia: a new case diagnosed during neonatal screening with tandem mass spectrometry. <i>Rapid Communications in Mass Spectrometry</i> , 2005, 19, 863-864.	0.7	9
103	A Pharmacokinetic Study and Correlation with Clinical Response of Rufinamide in Infants with Epileptic Encephalopathies. <i>Pharmacology</i> , 2013, 91, 275-280.	0.9	9
104	Bone status of children born from mothers with autoimmune diseases treated during pregnancy with prednisone and/or low molecular weight heparin. <i>Pediatric Rheumatology</i> , 2014, 12, 47.	0.9	9
105	A successful unrelated peripheral blood stem cell transplantation with reduced intensity conditioning regimen in a patient with late-onset purine nucleoside phosphorylase deficiency. <i>Pediatric Transplantation</i> , 2015, 19, E47-50.	0.5	9
106	Study protocol: safety and efficacy of propranolol 0.2% eye drops in newborns with a precocious stage of retinopathy of prematurity (DROP-ROP-0.2%): a multicenter, open-label, single arm, phase II trial. <i>BMC Pediatrics</i> , 2017, 17, 165.	0.7	9
107	Orange-colored diapers as first sign of Lesch-Nyhan disease in an asymptomatic infant. <i>Pediatric Nephrology</i> , 2010, 25, 2373-2374.	0.9	7
108	Late-Onset N-Acetylglutamate Synthase Deficiency: Report of a Paradigmatic Adult Case Presenting with Headaches and Review of the Literature. <i>International Journal of Molecular Sciences</i> , 2018, 19, 345.	1.8	7

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109	Effect of Beta 3 Adrenoreceptor Modulation on Patency of the Ductus Arteriosus. <i>Cells</i> , 2020, 9, 2625.	1.8	7
110	Inter-laboratory analytical improvement of succinylacetone and nitisinone quantification from dried blood spot samples. <i>JIMD Reports</i> , 2020, 53, 90-102.	0.7	7
111	Morquio B disease: From pathophysiology towards diagnosis. <i>Molecular Genetics and Metabolism</i> , 2021, 132, 180-188.	0.5	7
112	Multicenter evaluation of use of dried blood spot compared to conventional plasma in measurements of globotriaosylsphingosine (LysoGb3) concentration in 104 Fabry patients. <i>Clinical Chemistry and Laboratory Medicine</i> , 2021, 59, 1516-1526.	1.4	7
113	The successful inclusion of ADA SCID in Tuscany expanded newborn screening program. <i>Clinical Chemistry and Laboratory Medicine</i> , 2021, 59, e401-e404.	1.4	7
114	The diagnostic challenge of mild citrulline elevation at newborn screening. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 327-332.	0.5	7
115	First prenatal molecular diagnosis in a family with holocarboxylase synthetase deficiency. <i>Prenatal Diagnosis</i> , 2005, 25, 1117-1119.	1.1	6
116	Hypothermia for neonatal hypoxic-ischemic encephalopathy: may an early amplitude-integrated EEG improve the selection of candidates for cooling?. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2012, 25, 2171-2176.	0.7	6
117	PRDX1 gene-related epi-cblC disease is a common type of inborn error of cobalamin metabolism with mono- or bi-allelic MMACHC epimutations. <i>Clinical Epigenetics</i> , 2021, 13, 137.	1.8	6
118	Screening of Lysosomal Storage Disorders: Application of the Online Trapping-and-Cleanup Liquid Chromatography/Mass Spectrometry Method for Mucopolysaccharidosis I. <i>European Journal of Mass Spectrometry</i> , 2013, 19, 497-503.	0.5	5
119	Medium-Chain Acyl-CoA Deficiency: Outlines from Newborn Screening, <i>In Silico</i> Predictions, and Molecular Studies. <i>Scientific World Journal</i> , The, 2013, 2013, 1-8.	0.8	5
120	Upfront Enzyme Replacement via Erythrocyte Transfusions for PNP Deficiency. <i>Journal of Clinical Immunology</i> , 2021, 41, 1112-1115.	2.0	5
121	Liver-Directed Adeno-Associated Virus-Mediated Gene Therapy for Mucopolysaccharidosis Type VI. , 2022, 1, .		5
122	Biotinidase deficiency due to a de novo mutation or gonadal mosaicism in a first child. <i>Clinica Chimica Acta</i> , 2015, 445, 70-72.	0.5	4
123	Reducing the False-Positive Rate for Isovalerylcarnitine in Expanded Newborn Screening. <i>FIRE Forum for International Research in Education</i> , 2016, 4, 232640981666135.	0.7	4
124	Development and validation of a 2nd tier test for identification of purine nucleoside phosphorylase deficiency patients during expanded newborn screening by liquid chromatography-tandem mass spectrometry. <i>Clinical Chemistry and Laboratory Medicine</i> , 2016, 54, 627-32.	1.4	4
125	Development of a fast LC-MS/MS protocol for combined measurement of six LSDs on dried blood spot in a newborn screening program. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2019, 165, 135-140.	1.4	4
126	Study protocol: treatment with caffeine of the very preterm infant in the delivery room: a feasibility study. <i>BMJ Open</i> , 2020, 10, e040105.	0.8	4



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127	A new strategy implementing mass spectrometry in the diagnosis of congenital disorders of N-glycosylation (CDG). <i>Clinical Chemistry and Laboratory Medicine</i> , 2021, 59, 165-171.	1.4	4
128	Incessant Automatic Atrial Tachycardia in a Neonate Successfully Treated with Nadolol and Closely Spaced Doses of Flecainide: A Case Report. <i>Pediatric Reports</i> , 2020, 12, 108-113.	0.5	3
129	Serum creatinine during physiological perinatal dehydration may estimate individual nephron endowment. <i>European Journal of Pediatrics</i> , 2018, 177, 1383-1388.	1.3	2
130	Children with special health care needs attending emergency department in Italy: analysis of 3479 cases. <i>Italian Journal of Pediatrics</i> , 2020, 46, 173.	1.0	2
131	Lysosomals. , 2014, , 785-793.		2
132	Early Diagnosis and Treatment of Purine Nucleoside Phosphorylase (PNP) Deficiency through TREC-Based Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2021, 7, 62.	1.2	2
133	Metabolite and thymocyte development defects in ADA-SCID mice receiving enzyme replacement therapy. <i>Scientific Reports</i> , 2021, 11, 23221.	1.6	2
134	Newborn Screening: Are We Ready for It?. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, S10-S10.	1.1	0
135	Reticular dysgenesis-associated AK2 protects hematopoietic stem and progenitor cell development from oxidative stress. <i>Journal of Cell Biology</i> , 2015, 210, 210201A141.	2.3	0
136	Newborn Screening: Are We Ready for It?. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, S10.	1.1	0