Petr Chrastina

List of Publications by Year
in descending order

18	699	933447 10	996975 15
papers	citations	h-index	g-index

18	18	18	1124
all docs	docs citations	times ranked	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. Genetics in Medicine, 2011, 13, 230-254.	2.4	308
2	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117
3	Prosaposin deficiency and saposin B deficiency (activatorâ€deficient metachromatic leukodystrophy): Report on two patients detected by analysis of urinary sphingolipids and carrying novel PSAP gene mutations. American Journal of Medical Genetics, Part A, 2009, 149A, 613-621.	1.2	79
4	Determination of creatinine in urine by tandem mass spectrometry. Clinica Chimica Acta, 2004, 350, 99-106.	1.1	52
5	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	3.6	37
6	Simultaneous determination of cystathionine, total homocysteine, and methionine in dried blood spots by liquid chromatography/tandem mass spectrometry and its utility for the management of patients with homocystinuria. Clinica Chimica Acta, 2014, 437, 211-217.	1.1	32
7	Epidemiology of rare diseases detected by newborn screening in the Czech Republic. Central European Journal of Public Health, 2019, 27, 153-159.	1.1	16
8	Abnormalities in succinylpurines in fumarase deficiency: Possible role in pathogenesis of CNS impairment. Journal of Inherited Metabolic Disease, 2000, 23, 371-374.	3.6	15
9	Two novel mutations in mitochondrial acetoacetyl-CoA thiolase deficiency. Journal of Inherited Metabolic Disease, 2005, 28, 235-236.	3.6	13
10	Carnitine supplementation alleviates lipid metabolism derangements and protects against oxidative stress in non-obese hereditary hypertriglyceridemic rats. Applied Physiology, Nutrition and Metabolism, 2015, 40, 280-291.	1.9	13
11	Neonatal screening in the Czech Republic: increased prevalence of selected diseases in low birthweight neonates. European Journal of Pediatrics, 2018, 177, 1697-1704.	2.7	5
12	Impact of Newborn Screening and Early Dietary Management on Clinical Outcome of Patients with Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency and Medium Chain Acyl-CoA Dehydrogenase Deficiencyâ€"A Retrospective Nationwide Study. Nutrients, 2021, 13, 2925.	4.1	4
13	Extremely low birthweight neonates with phenylketonuria require special dietary management. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 2994-2999.	1.5	3
14	The role of <scp>ERNDIM</scp> Diagnostic Proficiency Schemes in improving the quality of diagnostic testing for Inherited Metabolic Diseases. Journal of Inherited Metabolic Disease, 2022, , .	3.6	3
15	Acylcarnitinesâ€™ Level in the Dried Blood Spot Samples of Healthy Newborns in Serbia-The Pilot Study. Serbian Journal of Experimental and Clinical Research, 2023, 24, 193-198.	0.1	1
16	Newborn screening for homocystinurias: recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 0, , .	3.6	1
17	X-linked adrenoleukodystrophy: phenotype-genotype correlation in hemizygous males and heterozygous females with ABCD1 mutations. Neuroendocrinology Letters, 2021, 42, 359-367.	0.2	0
18	The markers of the organic acidemias and their ratios in healthy neonates in Serbian population. Drug Metabolism and Personalized Therapy, 2022, .	0.6	0