Silvia Tortorelli

List of Publications by Year
in descending order

20	1,063	623734	752698
papers	citations	14 h-index	20 g-index

20	20	20	1274
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Immune dysfunction in MGAT2â€CDG : A clinical report and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 213-218.	1.2	5
2	The low excretor phenotype of glutaric acidemia type I is a source of false negative newborn screening results and challenging diagnoses. JIMD Reports, 2021, 60, 67-74.	1.5	4
3	A new ‹scp›Dâ€galactose‹/scp› treatment monitoring index for ‹scp›PGM1â€CDG‹/scp›. Journal of Inherited Metabolic Disease, 2021, 44, 1263-1271.	3.6	10
4	Biochemical Diagnosis of Acute Hepatic Porphyria: Updated Expert Recommendations for Primary Care Physicians. American Journal of the Medical Sciences, 2021, 362, 113-121.	1.1	24
5	Multiplex testing for the screening of lysosomal storage disease in urine: Sulfatides and glycosaminoglycan profiles in 40 cases of sulfatiduria. Molecular Genetics and Metabolism, 2020, 129, 106-110.	1.1	10
6	Biochemical phenotype and its relationship to treatment in 16 individuals with PCCB c.1606AÂ>ÂG (p.Asn536Asp) variant propionic acidemia. Molecular Genetics and Metabolism, 2020, 131, 316-324.	1.1	2
7	The Clinical Impact of CLIR Tools toward Rapid Resolution of Post-Newborn Screening Confirmatory Testing for X-Linked Adrenoleukodystrophy in California. International Journal of Neonatal Screening, 2020, 6, 62.	3.2	5
8	A Comparative Effectiveness Study of Newborn Screening Methods for Four Lysosomal Storage Disorders. International Journal of Neonatal Screening, 2020, 6, 44.	3.2	23
9	Incorporation of Second-Tier Biomarker Testing Improves the Specificity of Newborn Screening for Mucopolysaccharidosis Type I. International Journal of Neonatal Screening, 2020, 6, 10.	3.2	32
10	The critical role of psychosine in screening, diagnosis, and monitoring of Krabbe disease. Genetics in Medicine, 2020, 22, 1108-1118.	2.4	39
11	The Combined Impact of CLIR Post-Analytical Tools and Second Tier Testing on the Performance of Newborn Screening for Disorders of Propionate, Methionine, and Cobalamin Metabolism. International Journal of Neonatal Screening, 2020, 6, 33.	3.2	19
12	Acute liver failure in neonates with undiagnosed hereditary fructose intolerance due to exposure from widely available infant formulas. Molecular Genetics and Metabolism, 2018, 123, 428-432.	1.1	40
13	Precision newborn screening for lysosomal disorders. Genetics in Medicine, 2018, 20, 847-854.	2.4	99
14	Simultaneous Testing for 6 Lysosomal Storage Disorders and X-Adrenoleukodystrophy in Dried Blood Spots by Tandem Mass Spectrometry. Clinical Chemistry, 2016, 62, 1248-1254.	3.2	37
15	Continuous Age- and Sex-Adjusted Reference Intervals of Urinary Markers for Cerebral Creatine Deficiency Syndromes: A Novel Approach to the Definition of Reference Intervals. Clinical Chemistry, 2015, 61, 760-768.	3.2	38
16	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
17	Newborn screening for X-linked adrenoleukodystrophy (X-ALD): Validation of a combined liquid chromatographyâ€“tandem mass spectrometric (LCâ€“MS/MS) method. Molecular Genetics and Metabolism, 2009, 97, 212-220.	1.1	154
18	Combined liquid chromatographyâ€“Tandem mass spectrometry as an analytical method for high throughput screening for X-linked adrenoleukodystrophy and other peroxisomal disorders: Preliminary findings. Molecular Genetics and Metabolism, 2006, 89, 185-187.	1.1	97

#	Article	IF	Citations
19	Making the case for objective performance metrics in newborn screening by tandem mass spectrometry. Mental Retardation and Developmental Disabilities Research Reviews, 2006, 12, 255-261.	3.6	52
20	Recent developments and new applications of tandem mass spectrometry in newborn screening. Current Opinion in Pediatrics, 2004, 16, 427-433.	2.0	65