Rodolfo Tonin

List of Publications by Year
in descending order

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Version: 2024-02-01

14 papers	233 citations	1040056 9 h-index	1058476 14 g-index
15 all docs	15 docs citations	15 times ranked	332 citing authors

#	Article	IF	Citations
1	Galactosialidosis: review and analysis of CTSA gene mutations. Orphanet Journal of Rare Diseases, 2013, 8, 114.	2.7	48
2	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. BBA Clinical, 2016, 5, 114-119.	4.1	27
3	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. Human Mutation, 2015, 36, 357-368.	2.5	26
4	Type I sialidosis, a normosomatic lysosomal disease, in the differential diagnosis of late-onset ataxia and myoclonus: An overview. Molecular Genetics and Metabolism, 2020, 129, 47-58.	1.1	26
5	Carbon Nanotubes/Regenerated Silk Composite as a Three-Dimensional Printable Bio-Adhesive Ink with Self-Powering Properties. ACS Applied Materials & Interfaces, 2021, 13, 21007-21017.	8.0	17
6	3D Printing Silk-Based Bioresorbable Piezoelectric Self-Adhesive Holey Structures for In Vivo Monitoring on Soft Tissues. ACS Applied Materials & Interfaces, 2022, 14, 19253-19264.	8.0	15
7	Mis-splicing of the GALNS gene resulting from deep intronic mutations as a cause of Morquio a disease. BMC Medical Genetics, 2018, 19, 183.	2.1	14
8	Molecular basis of mucopolysaccharidosis IVA (Morquio A syndrome): A review and classification of GALNS gene variants and reporting of 68 novel variants. Human Mutation, 2021, 42, 1384-1398.	2.5	14
9	Pre-diagnosing and managing patients with GM1 gangliosidosis and related disorders by the evaluation of GM1 ganglioside content. Scientific Reports, 2019, 9, 17684.	3.3	11
10	High frequency of biotinidase deficiency in Italian population identified by newborn screening. Molecular Genetics and Metabolism Reports, 2020, 25, 100689.	1.1	10
11	Progressive myoclonus epilepsy in Gaucher Disease due to a new Glyâ€“Gly mutation causing loss of an Exonic Splicing Enhancer. Journal of Neurology, 2019, 266, 92-101.	3.6	9
12	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	1.1	7
13	Medium-Chain Acyl-CoA Deficiency: Outlines from Newborn Screening,In SilicoPredictions, and Molecular Studies. Scientific World Journal, The, 2013, 2013, 1-8.	2.1	5
14	Biotinidase deficiency due to a de novo mutation or gonadal mosaicism in a first child. Clinica Chimica Acta, 2015, 445, 70-72.	1.1	4