Rodolfo Tonin

List of Publications by Year
in descending order

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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	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
2	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	1.1	7
3	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
4	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
5	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
6	High frequency of biotinidase deficiency in Italian population identified by newborn screening. Molecular Genetics and Metabolism Reports, 2020, 25, 100689.	1.1	10
7	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
8	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
9	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
10	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
11	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
12	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
13	Galactosialidosis: review and analysis of CTSA gene mutations. Orphanet Journal of Rare Diseases, 2013, 8, 114.	2.7	48
14	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