Birute Tumiene

List of Publications by Year
in descending order

26	849	567281 15	610901 24
papers	citations	h-index	g-index

26	26	26	1746
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
2	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	3.6	146
3	Treatable inherited metabolic disorders causing intellectual disability: 2021 review and digital app. Orphanet Journal of Rare Diseases, 2021, 16, 170.	2.7	52
4	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
5	Diagnostic exome sequencing of syndromic epilepsy patients in clinical practice. Clinical Genetics, 2018, 93, 1057-1062.	2.0	39
6	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	6.2	35
7	Digitalisation and COVID-19: The Perfect Storm. Biomedicine Hub, 2020, 5, 1-23.	1.2	34
8	Metabolic Evaluation of Epilepsy: A Diagnostic Algorithm With Focus on Treatable Conditions. Frontiers in Neurology, 2018, 9, 1016.	2.4	33
9	Rare disease care pathways in the EU: from odysseys and labyrinths towards highways. Journal of Community Genetics, 2021, 12, 231-239.	1.2	32
10	Sleep and Behavioral Problems in Rolandic Epilepsy. Pediatric Neurology, 2013, 48, 115-122.	2.1	29
11	European Reference Networks: challenges and opportunities. Journal of Community Genetics, 2021, 12, 217-229.	1.2	21
12	The high frequency of GJB2 gene mutation c.313_326del14 suggests its possible origin in ancestors of Lithuanian population. BMC Genetics, 2016, 17, 45.	2.7	18
13	Nutritional and immune impairments and their effects on outcomes in early pancreatic cancer patients undergoing pancreatoduodenectomy. Clinical Nutrition, 2020, 39, 3385-3394.	5.0	18
14	Clinical and molecular characterization of a second case of 7p22.1 microduplication. American Journal of Medical Genetics, Part A, 2012, 158A, 1200-1203.	1.2	17
15	Phenotype comparison confirms ZMYND11 as a critical gene for 10p15.3 microdeletion syndrome. Journal of Applied Genetics, 2017, 58, 467-474.	1.9	17
16	A Scoping Review of Inborn Errors of Metabolism Causing Progressive Intellectual and Neurologic Deterioration (PIND). Frontiers in Neurology, 2019, 10, 1369.	2.4	16
17	Inflammatory myopathy in a patient with Aicardi-GoutiÃ¨res syndrome. European Journal of Medical Genetics, 2017, 60, 154-158.	1.3	14
18	Time for Change? The Why, What and How of Promoting Innovation to Tackle Rare Diseases â€“ Is It Time to Update the EUâ€™s Orphan Regulation? And if so, What Should be Changed?. Biomedicine Hub, 2020, 5, 1-11.	1.2	11

#	Article	IF	Citations
19	2022 Overview of Metabolic Epilepsies. Genes, 2022, 13, 508.	2.4	10
20	Contemporary scope of inborn errors of metabolism involving epilepsy or seizures. Metabolic Brain Disease, 2018, 33, 1781-1786.	2.9	9
21	Rare diseases: past achievements and future prospects. Journal of Community Genetics, 2021, 12, 205-206.	1.2	8
22	Multidisciplinary Care of Patients with Inherited Metabolic Diseases and Epilepsy: Current Perspectives. Journal of Multidisciplinary Healthcare, 2022, Volume 15, 553-566.	2.7	3
23	Features of KAT6B-related disorders in a patient with 10q22.1q22.3 deletion. Ophthalmic Genetics, 2017, 38, 383-386.	1.2	2
24	Unmet psychosocial needs of parents of children with rare, complex, and severe genetic diseases. Developmental Medicine and Child Neurology, 2021, , .	2.1	1
25	Imprinting and its disorders in evolutionary perspective. Acta Medica Lituanica, 2014, 21, 109-115.	0.3	0
26	Epileptogenic malformations of cortical development: when evolution goes awry. Acta Medica Lituanica, 2014, 21, 103-108.	0.3	0