

Sabine GrÃnberg

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

302
citations

1040056

9
h-index

996975

15
g-index

18
all docs

18
docs citations

18
times ranked

558
citing authors

#	ARTICLE	IF	CITATIONS
1	Genotype-phenotype correlations in <i>SCN8A</i> -related disorders reveal prognostic and therapeutic implications. <i>Brain</i> , 2022, 145, 2991-3009.	7.6	69
2	Phenotype and genotype in 103 patients with tricho-rhino-phalangeal syndrome. <i>European Journal of Medical Genetics</i> , 2015, 58, 279-292.	1.3	62
3	Clinical and biochemical improvement with galactose supplementation in <i>SLC35A2</i> -CDG. <i>Genetics in Medicine</i> , 2020, 22, 1102-1107.	2.4	42
4	Efficacy and safety of arimoclomol in Niemann-Pick disease type C: Results from a double-blind, randomised, placebo-controlled, multinational phase 2/3 trial of a novel treatment. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1463-1480.	3.6	26
5	Mortality and causes of death in children referred to a tertiary epilepsy center. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 66-71.	1.6	19
6	Diagnostic pitfalls in vitamin B6-dependent epilepsy caused by mutations in the <i>PLPBP</i> gene. <i>JIMD Reports</i> , 2019, 50, 1-8.	1.5	16
7	A Faroese founder variant in <i>TBCD</i> causes early onset, progressive encephalopathy with a homogenous clinical course. <i>European Journal of Human Genetics</i> , 2018, 26, 1512-1520.	2.8	13
8	Clinical disease progression and biomarkers in Niemann-Pick disease type C: a prospective cohort study. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 328.	2.7	12
9	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 350-356.	1.1	11
10	Modified Delphi procedure-based expert consensus on endpoints for an international disease registry for Metachromatic Leukodystrophy: The European Metachromatic Leukodystrophy initiative (MLDi). <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 48.	2.7	10
11	Monozygotic twins with a de novo 0.32 Mb 16q24.3 deletion, including <i>TUBB3</i> presenting with developmental delay and mild facial dysmorphism but without overt brain malformation. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2731-2736.	1.2	8
12	Case report: AARS2 leukodystrophy™. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 28, 100782.	1.1	7
13	Allogenic hematopoietic stem cell transplantation in two siblings with adult metachromatic leukodystrophy and a systematic literature review. <i>JIMD Reports</i> , 2021, 60, 96-104.	1.5	4
14	Expanding the cerebrovascular phenotype of the p.R258H variant in <i>ACTA2</i> related hereditary thoracic aortic disease (HTAD). <i>Journal of the Neurological Sciences</i> , 2020, 415, 116897.	0.6	2
15	Persistent Effect of Arimoclomol in Patients with Niemann-Pick Disease Type C: 24-Month Results from an Open-Label Extension of a Pivotal Phase 2/3 Study. <i>Neuropediatrics</i> , 2021, 52, .	0.6	0