Sabine GrÃ¸nborg

List of Publications by Year
in descending order

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 SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
2	Modified Delphi procedure-based expert consensus on endpoints for an international disease registry for Metachromatic Leukodystrophy: The European Metachromatic Leukodystrophy initiative (MLDi). Orphanet Journal of Rare Diseases, 2022, 17, 48.	2.7	10
3	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy. Molecular Genetics and Metabolism, 2022, 135, 350-356.	1.1	11
4	Allogenic hematopoietic stem cell transplantation in two siblings with adult metachromatic leukodystrophy and a systematic literature review. JIMD Reports, 2021, 60, 96-104.	1.5	4
5	Case report: â€˜AARS2 leukodystrophyâ€™. Molecular Genetics and Metabolism Reports, 2021, 28, 100782.	1.1	7
6	Efficacy and safety of arimoclomol in <scp>Niemannâ€Pick</scp> disease type C: Results from a doubleâ€blind, randomised, placeboâ€controlled, multinational phase 2/3 trial of a novel treatment. Journal of Inherited Metabolic Disease, 2021, 44, 1463-1480.	3.6	26
7	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. Neuropediatrics, 2021, 52, .	0.6	0
8	Clinical disease progression and biomarkers in Niemannâ€“Pick disease type C: a prospective cohort study. Orphanet Journal of Rare Diseases, 2020, 15, 328.	2.7	12
9	Expanding the cerebrovascular phenotype of the p.R258H variant in ACTA2 related hereditary thoracic aortic disease (HTAD). Journal of the Neurological Sciences, 2020, 415, 116897.	0.6	2
10	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. Genetics in Medicine, 2020, 22, 1102-1107.	2.4	42
11	Diagnostic pitfalls in vitamin B6â€dependent epilepsy caused by mutations in the PLPBP gene. JIMD Reports, 2019, 50, 1-8.	1.5	16
12	A Faroese founder variant in TBCD causes early onset, progressive encephalopathy with a homogenous clinical course. European Journal of Human Genetics, 2018, 26, 1512-1520.	2.8	13
13	Monozygotic twins with a de novo 0.32â€‰Mb 16q24.3 deletion, including TUBB3 presenting with developmental delay and mild facial dysmorphism but without overt brain malformation. American Journal of Medical Genetics, Part A, 2015, 167, 2731-2736.	1.2	8
14	Phenotype and genotype in 103 patients with tricho-rhino-phalangeal syndrome. European Journal of Medical Genetics, 2015, 58, 279-292.	1.3	62
15	Mortality and causes of death in children referred to a tertiary epilepsy center. European Journal of Paediatric Neurology, 2014, 18, 66-71.	1.6	19