Annette Bley

List of Publications by Year in descending order

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1478505 1588992 7 165 8 6 citations h-index g-index papers 8 8 8 338 docs citations times ranked citing authors all docs

#	ARTICLE	IF	CITATIONS
1	<i>PIGN</i> encephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
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	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in <i>POLR3A</i> , <i>POLR3B</i> , and <i>POLR1C</i> , Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
4	The natural history of Canavan disease: 23 new cases and comparison with patients from literature. Orphanet Journal of Rare Diseases, 2021, 16, 227.	2.7	9
5	POLR3A variants with striatal involvement and extrapyramidal movement disorder. Neurogenetics, 2020, 21, 121-133.	1.4	24
6	Ketogenic diet ameliorates axonal defects and promotes myelination in Pelizaeus–Merzbacher disease. Acta Neuropathologica, 2019, 138, 147-161.	7.7	48
7	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38