Rosalina M L Van Spaendonk

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	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
2	POLR3A variants with striatal involvement and extrapyramidal movement disorder. Neurogenetics, 2020, 21, 121-133.	1.4	24
3	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic <i>POLR1C</i> pathogenic variants. Neurology: Genetics, 2019, 5, e369.	1.9	38
4	Large exonic deletions in POLRB gene cause POLR3-related leukodystrophy. Orphanet Journal of Rare Diseases, 2015, 10, 69.	2.7	18
5	POLR3A and POLR3B Mutations in Unclassified Hypomyelination. Neuropediatrics, 2015, 46, 221-228.	0.6	22
6	Clinical spectrum of 4H leukodystrophy caused by <i>POLR3A</i> and <i>POLR3B</i> mutations. Neurology, 2014, 83, 1898-1905.	1.1	170