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Phenotypic and genetic spectrum of epilepsy with
myoclonic atonic seizures

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#	Paper	IF	Citations
23	Epilepsy With Myoclonic Atonic Seizures: Why Is the Yield of Genetic Testing for a "Presumed Genetic" Epilepsy Low?. <i>Epilepsy Currents</i> , 2020 , 20, 351-352	1.3	1
22	Complex Mosaicism of Two Distinct Mutations in a Female Patient With -Related Encephalopathy: A Case Report. <i>Frontiers in Genetics</i> , 2020 , 11, 911	4.5	2
21	Epilepsy with myoclonic-atic seizures (Dooze syndrome): Clarification of diagnosis and treatment options through a large retrospective multicenter cohort. <i>Epilepsia</i> , 2021 , 62, 120-127	6.4	9
20	Results of an international Delphi consensus in epilepsy with myoclonic atonic seizures/ Dooze syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021 , 85, 12-18	3.2	4
19	Deep-Phenotyping the Less Severe Spectrum of Deficiency and Linking the Gene to Myoclonic Atonic Seizures. <i>Frontiers in Genetics</i> , 2021 , 12, 663643	4.5	2
18	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies including Myoclonic Atonic Epilepsy.		
17	Further evidence for de novo variants in SYNCIP as the cause of a neurodevelopmental disorder. <i>Human Mutation</i> , 2021 , 42, 1094-1100	4.7	2
16	ZMYND11 variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. <i>Clinical Genetics</i> , 2021 , 100, 412-429	4	0
15	How We Got to Where WeVe Going. 2021 ,		0
14	Clinical next generation sequencing in developmental and epileptic encephalopathies: Diagnostic relevance of data re-analysis and variants re-interpretation. <i>European Journal of Medical Genetics</i> , 2021 , 64, 104363	2.6	1
13	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. <i>Brain</i> , 2021 ,	11.2	2
12	Deficiency of autism risk factor ASH1L in prefrontal cortex induces epigenetic aberrations and seizures. <i>Nature Communications</i> , 2021 , 12, 6589	17.4	6
11	Epilepsy with myoclonic-atic seizures, also known as Dooze syndrome: Modification of the diagnostic criteria. <i>European Journal of Paediatric Neurology</i> , 2021 , 36, 37-50	3.8	1
10	Epileptic Phenotypes Associated With SNAREs and Related Synaptic Vesicle Exocytosis Machinery.. <i>Frontiers in Neurology</i> , 2021 , 12, 806506	4.1	0
9	Myoclonic Epilepsy: Case Report of a Mild Phenotype in a Pediatric Patient Expanding Clinical Spectrum of Pathogenic Variants.. <i>Frontiers in Neurology</i> , 2021 , 12, 806516	4.1	
8	Synaptopathies in Developmental and Epileptic Encephalopathies: A Focus on Pre-synaptic Dysfunction.. <i>Frontiers in Neurology</i> , 2022 , 13, 826211	4.1	1
7	Molecular and Clinical Repercussions of GABA Transporter 1 Variants Gone Amiss: Links to Epilepsy and Developmental Spectrum Disorders.. <i>Frontiers in Molecular Biosciences</i> , 2022 , 9, 834498	5.6	2

6	Genetic Epilepsy Syndromes.. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2022 , 28, 339-362	3	
5	Nomenclature of Genetic Movement Disorders: Recommendations of the International Parkinson and Movement Disorder Society Task Force - An Update.. <i>Movement Disorders</i> , 2022 , 37, 905-935	7	3
4	Use of sulthiame as add-on therapy in children with myoclonic atonic epilepsy: A study of 35 patients.. <i>Epilepsy and Behavior</i> , 2022 , 131, 108702	3.2	
3	Myoclonic-Atonic Epilepsy Caused by a Novel de Novo Heterozygous Missense Variant in the SLC6A1 Gene: Brief Discussion of the Literature and Detailed Case Description of a Severely Intellectually Disabled Adult Male Patient. Volume 15, 753-759		o
2	A draft conceptual model of SLC6A1 neurodevelopmental disorder. 16,		o
1	The role of histone methyltransferases in neurocognitive disorders associated with brain size abnormalities. 17,		o