

Diagnosis of neuronal ceroid lipofuscinosis type 2 (CLN2) for early detection and laboratory diagnosis

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Management Strategies for CLN2 Disease. <i>Pediatric Neurology</i> , 2017, 69, 102-112.	1.0	80
2	Photosensitivity is an early marker of neuronal ceroid lipofuscinosis type 2 disease. <i>Epilepsia</i> , 2017, 58, 1380-1388.	2.6	50
3	Translating preclinical models of neuronal ceroid lipofuscinosis: progress and prospects. <i>Expert Opinion on Orphan Drugs</i> , 2017, 5, 727-740.	0.5	6
4	Gene Therapy Approaches to Treat the Neurodegeneration and Visual Failure in Neuronal Ceroid Lipofuscinoses. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1074, 91-99.	0.8	14
5	Evolution and course of early life developmental encephalopathic epilepsies: Focus on Lennox-Gastaut syndrome. <i>Epilepsia</i> , 2018, 59, 2096-2105.	2.6	35
6	Metabolic ataxias. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 155, 117-127.	1.0	5
7	Mutation update: Review of <i>TPP1</i> gene variants associated with neuronal ceroid lipofuscinosis CLN2 disease. <i>Human Mutation</i> , 2019, 40, 1924-1938.	1.1	46
8	Advances in the treatment of neuronal ceroid lipofuscinosis. <i>Expert Opinion on Orphan Drugs</i> , 2019, 7, 473-500.	0.5	20
9	Perampanel attenuates myoclonus in a patient with neuronal ceroid lipofuscinoses type 2 disease. <i>Brain and Development</i> , 2019, 41, 817-819.	0.6	8
10	Global Brain Transcriptome Analysis of a <i>Tpp1</i> Neuronal Ceroid Lipofuscinoses Mouse Model. <i>ASN Neuro</i> , 2019, 11, 175909141984339.	1.5	13
11	Validity of a rapid and simple fluorometric tripeptidyl peptidase 1 (TPP1) assay using dried blood specimens to diagnose CLN2 disease. <i>Clinica Chimica Acta</i> , 2019, 492, 69-71.	0.5	14
12	Therapeutic landscape for Batten disease: current treatments and future prospects. <i>Nature Reviews Neurology</i> , 2019, 15, 161-178.	4.9	127
13	Overview of advances in educational and social supports for young persons with NCL disorders. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2020, 1866, 165480.	1.8	2
14	Autism, Epilepsy, and Neuroregression: Photosensitivity on Electroencephalography Solved the Riddle. <i>Clinical EEG and Neuroscience</i> , 2020, 51, 399-402.	0.9	6
15	Review of Cerliponase Alfa: Recombinant Human Enzyme Replacement Therapy for Late-Infantile Neuronal Ceroid Lipofuscinosis Type 2. <i>Journal of Child Neurology</i> , 2020, 35, 348-353.	0.7	20
16	Slowing late infantile Batten disease by direct brain parenchymal administration of a rh.10 adeno-associated virus expressing <i>CLN2</i> . <i>Science Translational Medicine</i> , 2020, 12, .	5.8	35
17	Cerliponase alfa for CLN2 disease, a promising therapy. <i>Expert Opinion on Orphan Drugs</i> , 2020, 8, 445-454.	0.5	1
18	The improvement in diagnosis and epilepsy managing in children with progressive myoclonus epilepsy during the last decade – A tertiary center experience in cohort of 51 patients. <i>Epilepsy and Behavior</i> , 2020, 113, 107456.	0.9	5

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19	Intracerebroventricular Cerliponase Alfa for Neuronal Ceroid Lipofuscinosis Type 2 Disease: Clinical Practice Considerations From US Clinics. <i>Pediatric Neurology</i> , 2020, 110, 64-70.	1.0	12
20	A multimodal approach to identify clinically relevant biomarkers to comprehensively monitor disease progression in a mouse model of pediatric neurodegenerative disease. <i>Progress in Neurobiology</i> , 2020, 189, 101789.	2.8	9
21	Neuronal ceroid lipofuscinosis in the Russian population: Two novel mutations and the prevalence of heterozygous carriers. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1228.	0.6	9
22	Targeted re-sequencing for early diagnosis of genetic causes of childhood epilepsy: the Italian experience from the "beyond epilepsy" project. <i>Italian Journal of Pediatrics</i> , 2020, 46, 92.	1.0	17
23	Drug Treatment of Progressive Myoclonic Epilepsy. <i>Paediatric Drugs</i> , 2020, 22, 149-164.	1.3	12
24	Neuronal ceroid lipofuscinosis type 2: an Australian case series. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 1210-1218.	0.4	19
25	Diagnosis of late-infantile neuronal ceroid lipofuscinosis using dried blood spot-based assay for TPP1 enzyme activity. <i>Clinica Chimica Acta</i> , 2020, 507, 62-68.	0.5	1
26	<p><p>Changing Times for CLN2 Disease: The Era of Enzyme Replacement Therapy<p><p>. <i>Therapeutics and Clinical Risk Management</i> , 2020, Volume 16, 213-222.	0.9	34
27	Neuronal Ceroid Lipofuscinosis: Potential for Targeted Therapy. <i>Drugs</i> , 2021, 81, 101-123.	4.9	35
28	Cerliponase alfa changes the natural history of children with neuronal ceroid lipofuscinosis type 2: The first French cohort. <i>European Journal of Paediatric Neurology</i> , 2021, 30, 17-21.	0.7	13
29	"Atypical" Phenotypes of Neuronal Ceroid Lipofuscinosis: The Argentine Experience in the Genomic Era. <i>Journal of Inborn Errors of Metabolism and Screening</i> , 0, 9, .	0.3	1
30	Managing CLN2 disease: a treatable neurodegenerative condition among other treatable early childhood epilepsies. <i>Expert Review of Neurotherapeutics</i> , 2021, 21, 1275-1282.	1.4	5
31	Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 185.	1.2	17
32	"Real world effectiveness of cerliponase alfa in classical and atypical patients. A case series". <i>Molecular Genetics and Metabolism Reports</i> , 2021, 27, 100718.	0.4	7
33	Ocular Manifestations of Neuronal Ceroid Lipofuscinoses. <i>Seminars in Ophthalmology</i> , 2021, 36, 582-595.	0.8	2
34	Unraveling neuronal ceroid lipofuscinosis type 2 (CLN2) disease: A tertiary center experience for determinants of diagnostic delay. <i>European Journal of Paediatric Neurology</i> , 2021, 33, 94-98.	0.7	4
35	An ERG and OCT study of neuronal ceroid lipofuscinosis CLN2 Battens retinopathy. <i>Eye</i> , 2021, 35, 2438-2448.	1.1	6
36	A diagnostic confidence scheme for <sc>CLN3</sc> disease. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1453-1462.	1.7	3

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38	Revealing the clinical phenotype of atypical neuronal ceroid lipofuscinosis type 2 disease: Insights from the largest cohort in the world. <i>Journal of Paediatrics and Child Health</i> , 2021, 57, 519-525.	0.4	15
39	New Advanced Strategies for the Treatment of Lysosomal Diseases Affecting the Central Nervous System. <i>Current Pharmaceutical Design</i> , 2019, 25, 1933-1950.	0.9	11
40	Patient-Derived Induced Pluripotent Stem Cell Models for Phenotypic Screening in the Neuronal Ceroid Lipofuscinoses. <i>Molecules</i> , 2021, 26, 6235.	1.7	4
41	Praise for child neuropsychiatry. <i>Turk Pediatri Arsivi</i> , 2019, 55, 453-454.	0.9	0
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44	A Case Report on the Challenging Diagnosis of Neuronal Ceroid Lipofuscinosis Type 2 (CLN2). <i>Journal of Inborn Errors of Metabolism and Screening</i> , 0, 8, .	0.3	1
45	Is it autism? Some suggestions for pediatricians. <i>Turk Pediatri Arsivi</i> , 2020, 55, 229-235.	0.9	7
46	Position of Experts Regarding Follow-Up of Patients with Neuronal Ceroid Lipofuscinosis-2 Disease in Latin America. <i>Journal of Inborn Errors of Metabolism and Screening</i> , 0, 8, .	0.3	2
47	Brain transcriptome analysis of a CLN2 mouse model as a function of disease progression. <i>Journal of Neuroinflammation</i> , 2021, 18, 262.	3.1	5
48	Neurophysiological Findings in Neuronal Ceroid Lipofuscinoses. <i>Frontiers in Neurology</i> , 2022, 13, 845877.	1.1	7
49	The LINCE Project: A Pathway for Diagnosing NCL2 Disease. <i>Frontiers in Pediatrics</i> , 2022, 10, 876688.	0.9	1
51	Value of genetic testing for pediatric epilepsy: Driving earlier diagnosis of ceroid lipofuscinosis type 2 Batten disease. <i>Epilepsia</i> , 2022, 63, .	2.6	6
52	Recent Insight into the Genetic Basis, Clinical Features, and Diagnostic Methods for Neuronal Ceroid Lipofuscinosis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 5729.	1.8	7
53	Neuronal Ceroid Lipofuscinosis Type 2: A Case Series from Argentina. <i>Journal of Inborn Errors of Metabolism and Screening</i> , 0, 10, .	0.3	0
54	Provoked seizures at the onset of progressive disease contribute to diagnosis delay - A tertiary center experience in a cohort of 22 children with CLN2. <i>European Journal of Paediatric Neurology</i> , 2022, 40, 1-4.	0.7	1
55	Role of Electroencephalogram (EEG) and Magnetic Resonance Imaging (MRI) Findings in Early Recognition and Diagnosis of Neuronal Ceroid Lipofuscinosis Type 2 Disease. <i>Journal of Child Neurology</i> , 2022, 37, 984-991.	0.7	1
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58	Cynomolgus macaque model of neuronal ceroid lipofuscinosis type 2 disease. <i>Experimental Neurology</i> , 2023, 363, 114381.	2.0	0
59	The conserved cellular roles of CLN proteins: Novel insights from <i>Dictyostelium discoideum</i> . <i>European Journal of Cell Biology</i> , 2023, 102, 151305.	1.6	1
61	Optimizing Therapy of Seizures in Children and Adolescents with Developmental Disabilities. , 2023, , 631-653.		0
62	Economic analysis of cerliponase alfa for treatment of late-infantile neuronal ceroid lipofuscinosis type 2 (CLN2). <i>Expert Review of Pharmacoeconomics and Outcomes Research</i> , 2023, 23, 561-570.	0.7	1
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