CLN2 Disease Selected Publications

CLN2 Disease and Diagnosis	Leal-Pardinas F, Truty R, McKnight DA, et al. Value of genetic testing for pediatric epilepsy: driving earlier diagnosis of ceroid lipofuscinosis type 2 Batten disease. *Epilepsia*. 2022;63(7):e68-e73. DOI: 10.1111/epi.17269
	Lourenço CM, Pessoa A, Mendes CC, et al. Revealing the clinical phenotype of atypical neuronal ceroid lipofuscinosis type 2 disease: Insights from the largest cohort in the world. *J Paediatr Child Health*. 2021;57(4):519–525. DOI: 10.1111/jpc.15250
	Gardner E, Bailey M, Schulz A, et al. Mutation update: Review of TPP1 gene variants associated with neuronal ceroid lipofuscinosis CLN2 disease. *Hum Mutat*. 2019;40(11):1924-1938. DOI: 10.1002/humu.23860
	Nickel M, Simonati A, Jacoby D, et al. Disease characteristics and progression in patients with late-infantile neuronal ceroid lipofuscinosis type 2 (CLN2) disease: An observational cohort study. *Lancet Child Adolesc Health*. 2018;2(8):582–590. DOI: 10.1016/S2352-4642(18)30179-2[a]
	Fietz M, Al Sayed M, Burke D, et al. Diagnosis of neuronal ceroid lipofuscinosis type 2 (CLN2 disease): Expert recommendations for early detection and laboratory diagnosis. *Mol Genet Metab.*2016;119(1–2):160–167. DOI: 10.1016/j.ymgme.2016.07.011
Cerliponase alfa	Schulz A, Schwering C, Wibbeler E, et al. Real-world clinical outcomes of patients with CLN2 disease treated with cerliponase alfa. *Front Neurol*. 2025; 16:1516026. DOI: 10.3389/fneur.2025.1516026
	Schulz A, Specchio N, de Los Reyes E, et al. Safety and efficacy of cerliponase alfa in children with neuronal ceroid lipofuscinosis type 2 (CLN2 disease): an open-label extension study. *Lancet Neurol*. 2024;23(1):60-70. DOI: 10.1016/S1474-4422(23)00384-8[a]
	Schwering C, Kammler G, Wibbeler E, et al. Development of the "Hamburg Best Practice Guidelines for ICV-Enzyme Replacement Therapy (ERT) in CLN2 Disease" based on 6 years treatment experience in 48 patients. *J Child Neurol.* 2021;36(8):635–641. DOI: 10.1177/08830738219891
	Wibbeler E, Wang R, Reyes EL, et al. Cerliponase alfa for the treatment of atypical phenotypes of CLN2 disease: A retrospective case series. *J Child Neurol*. 2021;36(6):468-474. DOI: 10.1177/0883073820977997
	de Los Reyes E, Lehwald L, Augustine EF, et al. Intracerebroventricular cerliponase alfa for neuronal ceroid lipofuscinosis type 2 disease: Clinical practice considerations from US clinics. *Pediatr Neurol*. 2020;110:64-70. DOI: 10.1016/j.pediatrneurol.2020.04.018
	Schulz A, Ajayi T, Specchio N, et al. Study of intraventricular cerliponase alfa for CLN2 disease. *N Engl J Med*. 2018;378(20):1898–1907. DOI: 10.1056/NEJMoa1712649
Management Guidelines	Mole SE, Schulz A, Badoe E, et al. Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. *Orphanet J Rare Dis*. 2021;16(1):185. DOI: 10.1186/s13023-021-01813-5
	Williams RE, Adams HR, Blohm M, et al. Management strategies for CLN2 disease. *Pediatr Neurol.* 2017;69:102–112. DOI: 10.1016/j.pediatrneurol.2017.01.034

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