

NCL2TREAT publication list

Cerliponase Alfa for the treatment of atypical phenotypes of CLN2 disease: A retrospective case series

J Child Neurol. Mai 2021. >>[PubMed-Link](#)<<

Wibbeler E, Wang R, de los Reyes E, Specchio N, Gissen P, Guelbert N, Nickel M, Schwering C, Lehwald L, Trivisano M, Lee L, Amato G, Cohen-Pfeffer J, Shediak R, Leal-Pardinas F, Schulz A.

Enzymatic diagnosis of neuronal lipofuscinoses in dried blood spots using substrates for concomitant tandem mass spectrometry and fluorimetry

J Mass Spectrom, Jan 2021, >>[PubMed-Link](#)<<

Maeser S, Petre BA, Ion L, Rawer S, Kohlschütter A, Santorelli FM, Simonati A, Schulz A, Przybylski M.

An ophthalmic rating scale to assess ocular involvement in juvenile CLN3

Am J Ophthalmol, Dez 2020 >>[PubMed-Link](#)<<

Dulz S, Atiskova Y, Wibbeler E, Wildner J, Wagenfeld L, Schwering C, Nickel M, Bartsch U, Spitzer MS, Schulz A.

Enzyme replacement therapy with recombinant pro-CTSD (cathepsin D) corrects defective proteolysis and autophagy in neuronal ceroid lipofuscinosis

Autophagy. 2019 Jul 16:1-15. doi: 10.1080/15548627.2019.1637200. [Epub ahead of print]

Marques ARA, Di Spiezio A, Thießen N, Schmidt L, Grötzinger J, Lüllmann-Rauch R, Damme M, Storck SE, Pietrzik CU, Fogh J, Bär J, Mikhaylova M, Glatzel M, Bassal M, Bartsch U, Saftig P

Lysosomal proteome analysis reveals that CLN3-defective cells have multiple enzyme deficiencies associated with changes in intracellular trafficking

J Biol Chem. 2019 Apr 30. pii: jbc.RA119.008852. doi: 10.1074/jbc.RA119.008852. [Epub ahead of print]

Schmidtke C, Tiede S, Thelen M, Käkelä R, Jabs S, Makrypidi G, Sylvester M, Schweizer M6, Braren I, Brocke-Ahmadinejad N, Cotman SL, Schulz A, Gieselmann V, Bräulke T

Disease characteristics and progression in patients with late-infantile neuronal ceroid lipofuscinosis type 2 (CLN2) disease: an observational cohort study.

Lancet Child Adolesc Health. 2(8):582-590

Nickel M, Simonati A, Jacoby D, Lezius S, Kilian D, Van de Graaf B, Pagovich OE, Kosofsky B, Yohay K, Downs M, Slasor P, Ajayi T, Crystal RG, Kohlschütter A, Sondhi D, Schulz A.

Study of Intraventricular Cerliponase Alfa for CLN2 Disease

N Engl J Med 2018 May 17; 378:1898-1907 DOI: 10.1056/NEJMoa1712649N

Schulz A, Ajayi T, Specchio N, de Los Reyes E, Gissen P, Ballon D, Dyke JP, Cahan H, Slasor P, Jacoby D, Kohlschütter A; CLN2 Study Group.

Volumetric Description of Brain Atrophy in Neuronal Ceroid Lipofuscinosis 2: Supratentorial Gray Matter Shows Uniform Disease Progression.

AJNR Am J Neuroradiol. 2016 May 26.

Löbel U, Sedlacik J, Nickel M, Lezius S, Fiehler J, Nestrasil I, Kohlschütter A, Schulz A.