

Resource Summary Report

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NCL Resource - A gateway for Batten disease

RRID:SCR_012826

Type: Tool

Proper Citation

NCL Resource - A gateway for Batten disease (RRID:SCR_012826)

Resource Information

URL: <http://www.ucl.ac.uk/ncl/>

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Description: It serves as a gateway for clinicians, families and researchers who have an interest in or are affected by Batten disease or who wish to find out more. Information can be accessed via four main routes - Clinicians, Families, Researchers, Professional Support. The Clinical route describes Batten disease and includes details on diagnosis and diagnostic services. The Family route also describes Batten disease and lists support groups. The Research route includes the NCL Mutation Database, established in 1998, and other useful information. The Professional Support route includes details of coordinated initiatives to support those affected by Batten disease. A fifth route, Research Consortia, serves to meet research needs and currently act as a focus for collaborative efforts to identify the remaining human and animal NCL genes and facilitate functional approaches. An additional route, Creativity, has been launched to display creative items from families with Batten disease, and to celebrate life, in both its fullness and fragility.

Synonyms: NCL Resource

Resource Type: database, data or information resource

Keywords: FASEB list

Resource Name: NCL Resource - A gateway for Batten disease

Resource ID: SCR_012826

Alternate IDs: nif-0000-03182

Record Creation Time: 20220129T080312+0000

Record Last Update: 20260803T040614+0000

Ratings and Alerts

No rating or validation information has been found for NCL Resource - A gateway for Batten disease.

No alerts have been found for NCL Resource - A gateway for Batten disease.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 30 mentions in open access literature.

Listed below are recent publications. The full list is available at [ASWG](#) .

Luo S, et al. (2020) Functional Analysis of a Novel CLN5 Mutation Identified in a Patient With Neuronal Ceroid Lipofuscinosis. *Frontiers in genetics*, 11, 536221.

Eaton SL, et al. (2019) CRISPR/Cas9 mediated generation of an ovine model for infantile neuronal ceroid lipofuscinosis (CLN1 disease). *Scientific reports*, 9(1), 9891.

Jilani A, et al. (2019) High diagnostic yield of direct Sanger sequencing in the diagnosis of neuronal ceroid lipofuscinoses. *JIMD reports*, 50(1), 20.

di Ronza A, et al. (2018) CLN8 is an endoplasmic reticulum cargo receptor that regulates lysosome biogenesis. *Nature cell biology*, 20(12), 1370.

Gao Z, et al. (2018) Identification of two novel null variants in CLN8 by targeted next-generation sequencing: first report of a Chinese patient with neuronal ceroid lipofuscinosis due to CLN8 variants. *BMC medical genetics*, 19(1), 21.

Sheth J, et al. (2018) Batten disease: biochemical and molecular characterization revealing novel PPT1 and TPP1 gene mutations in Indian patients. *BMC neurology*, 18(1), 203.

Mitchell NL, et al. (2018) Longitudinal In Vivo Monitoring of the CNS Demonstrates the Efficacy of Gene Therapy in a Sheep Model of CLN5 Batten Disease. *Molecular therapy : the journal of the American Society of Gene Therapy*, 26(10), 2366.

Uusi-Rauva K, et al. (2017) Induced Pluripotent Stem Cells Derived from a CLN5 Patient Manifest Phenotypic Characteristics of Neuronal Ceroid Lipofuscinoses. *International journal of molecular sciences*, 18(5).

Beesley C, et al. (2017) CLN8 disease caused by large genomic deletions. *Molecular genetics & genomic medicine*, 5(1), 85.

Kim K, et al. (2017) Safety and potential efficacy of gemfibrozil as a supportive treatment for children with late infantile neuronal ceroid lipofuscinosis and other lipid storage disorders. *Orphanet journal of rare diseases*, 12(1), 113.

Marotta D, et al. (2017) NCLs and ER: A stressful relationship. *Biochimica et biophysica acta. Molecular basis of disease*, 1863(6), 1273.

Scifo E, et al. (2015) Proteomic analysis of the palmitoyl protein thioesterase 1 interactome in SH-SY5Y human neuroblastoma cells. *Journal of proteomics*, 123, 42.

Amorim IS, et al. (2015) Molecular neuropathology of the synapse in sheep with CLN5 Batten disease. *Brain and behavior*, 5(11), e00401.

Hughes SM, et al. (2014) Inhibition of storage pathology in prenatal CLN5-deficient sheep neural cultures by lentiviral gene therapy. *Neurobiology of disease*, 62, 543.

Koch S, et al. (2013) Cathepsin D deficiency induces cytoskeletal changes and affects cell migration pathways in the brain. *Neurobiology of disease*, 50, 107.

Passantino R, et al. (2013) Identifying protein partners of CLN8, an ER-resident protein involved in neuronal ceroid lipofuscinosis. *Biochimica et biophysica acta*, 1833(3), 529.

Kay GW, et al. (2013) Chronic oral administration of minocycline to sheep with ovine CLN6 neuronal ceroid lipofuscinosis maintains pharmacological concentrations in the brain but does not suppress neuroinflammation or disease progression. *Journal of neuroinflammation*, 10, 97.

Schmiedt ML, et al. (2012) Cln5-deficiency in mice leads to microglial activation, defective myelination and changes in lipid metabolism. *Neurobiology of disease*, 46(1), 19.

Kay GW, et al. (2011) The specific loss of GnRH-positive neurons from the hypothalamus of sheep with CLN6 neuronal ceroid lipofuscinosis occurs without glial activation and has only minor effects on reproduction. *Neurobiology of disease*, 41(3), 614.

Benitez BA, et al. (2011) Exome-sequencing confirms DNAJC5 mutations as cause of adult neuronal ceroid-lipofuscinosis. *PloS one*, 6(11), e26741.