Resource Summary Report

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Mutation and Patient Database

RRID:SCR_018806
Type: Tool

Proper Citation

Mutation and Patient Database (RRID:SCR_018806)

Resource Information

URL: https://www.ucl.ac.uk/ncl-disease/mutation-and-patient-database

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Description: Collection of published mutations and sequence variations in genes that cause Neuronal Ceroid Lipofuscinoses together with unpublished data included with permission. There are two tables for each human NCL disease gene - Patient Datasheets list all published or reported patients and families, and Mutation Datasheets list all published or reported mutations, cross-referenced to patient table. Datasheets are available to view or download as excel files for off-site use to aid local needs or interests. Database follows mutation nomenclature recommendations of Human Genome Variation Society.

Synonyms: NCL Mutation Database, NCL Mutation and Patient Database

Resource Type: data or information resource, data set, database

Keywords: Mutation, gene mutation, sequence variations, gene, human NCL disease gene, patient datasheet, mutation datasheet, mutation nomenclature, human genome variation society, data

Related Condition: Neuronal Ceroid Lipofuscinoses, NCL, Batten disease

Availability: Free, Freely available

Resource Name: Mutation and Patient Database

Resource ID: SCR_018806

Record Creation Time: 20220129T080342+0000

Record Last Update: 20260919T195358+0000

Ratings and Alerts

No rating or validation information has been found for Mutation and Patient Database.

No alerts have been found for Mutation and Patient Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 5 mentions in open access literature.

Listed below are recent publications. The full list is available at nidm-terms .

Simonati A, et al. (2026) Age at onset and gene variants predict lifespan and disease duration in childhood neuronal ceroid lipofuscinoses. Developmental medicine and child neurology, 68(2), 276.

Pasquetti D, et al. (2023) Linear Diagnostic Procedure Elicited by Clinical Genetics and Validated by mRNA Analysis in Neuronal Ceroid Lipofuscinosis 7 Associated with a Novel Non-Canonical Splice Site Variant in MFSD8. Genes, 14(2).

Soldati C, et al. (2021) Repurposing of tamoxifen ameliorates CLN3 and CLN7 disease phenotype. EMBO molecular medicine, 13(10), e13742.

Huber RJ, et al. (2020) Mfsd8 localizes to endocytic compartments and influences the secretion of Cln5 and cathepsin D in Dictyostelium. Cellular signalling, 70, 109572.

Seifert C, et al. (2020) Modulation of Kv4.2/KChIP3 interaction by the ceroid lipofuscinosis neuronal 3 protein CLN3. The Journal of biological chemistry, 295(34), 12099.