Summary of the New Classification of the NCLs

Soluble lysosomal enzyme deficiencies

Gene Symbol	Protein	Diseases
CTSD CLN10	Cathepsin D	CLN10 disease, congenital CLN10 disease, late infantile CLN10 disease, juvenile CLN10 disease, adult
PPT1 CLN1	Palmitoyl Protein Thioesterase 1 (PPT1)	CLN1 disease, infantile CLN1 disease, late infantile CLN1 disease, juvenile CLN1 disease, adult
TPP1 CLN2	Tripeptidyl Peptidase 1 (TPP1)	CLN2 disease, late infantile CLN2 disease, juvenile
CTSF CLN13	Cathepsin F	CLN13 disease, adult Kufs type B

Non-enzyme deficiencies
(functions of identified proteins tend to be poorly understood currently)

Gene Symbol	Protein	Diseases
CLN3	Transmembrane protein	CLN3 disease, juvenile
CLN5	Soluble; lysosomal	CLN5 disease, late infantile CLN5 disease, juvenile CLN5 disease, adult
CLN6	Transmembrane protein; ER	CLN6 disease, late infantile CLN6 disease, adult Kufs type A
MFSD8 CLN7	Major facilitator superfamily domain-containing protein 8 Transmembrane protein; Endolysosomal transporter	CLN7 disease, late infantile
CLN8	Transmembrane protein; ER, ER-Golgi intermediate complex	CLN8 disease, late infantile CLN8 disease, EPMR

Adapted by the BDFA from Dr Sara Mole and Dr Ruth Williams, NCL2012 Abstract Book (2012)

DNAJC5 CLN4	Soluble cysteine string protein α	CLN4 disease, adult autosomal dominant
GRN CLN11	Progranulin	CLN11 disease, adult Heterozygous mutations cause frontotemporal lobar dementia
ATP13A2 CLN12	P-type ATPase	CLN12 disease, juvenile Mutations also cause Kufor-Rakeb syndrome
KCTD7 CLN14	Potassium channel Tetramerization domain-containing protein 7	CLN14 disease, infantile Mutation also causes progressive myoclonic epilepsy-3

Others
(those whose classification is uncertain because of incomplete diagnostic investigations; absence of a confirmed gene/mutation designation; or where the NCL is of a rare or minor mutation-specific phenotype)

Gene Symbol	Protein	Diseases
?	Mutations not yet defined in any gene	Congenital/infantile variants
?	Mutations not yet defined in any gene	Late infantile variants
?CLN9?	Mutations not yet defined in any gene	Juvenile variants
?	Mutations not yet defined in any gene	Late onset/adult variants including some adult Kufs type B
CLCN6	Mutations not yet found on both disease alleles in human disease	Chloride transport defect, adult onset
SGSH	Mutations usually cause MPSIIIA	Adult onset

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