Summary of new classification nomenclature of the NCLs

	Gene symbol	Protein	Diseases
Soluble lysosomal enzyme deficiencies	*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 generally poorly understood at the current time)	*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
	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 or absence of confirmed gene/mutation designation, or where NCL is a rare or minor mutation-specific phenotype	*?*	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

Diseases listed are those now described, and all are autosomal recessive unless noted. It is possible that further cases of later onset e.g. CLN2 disease, adult, or those with atypical progression, e.g. CLN3 disease, juvenile, will yet be recognised.