e Table 1. Genes with pathogenic variants in the LICE PME cohort ordered according to number of unrelated patients.

Gene	Inheritance	Syndrome(s)	Number	Paper
CSTB	Recessive	Unverricht-Lundborg disease (ULD)	54	2014
EPM2A	Recessive	Lafora disease	7*	2014
NHLRC1	Recessive	Lafora disease	22*	2014
CLN6	Recessive	Neuronal ceroid lipofuscinosis (NCL)	5	2014, 2021
NEU1	Recessive	Sialidosis I, II	5	2014, 2021
MT-TK	Mitochondrial	Myoclonic epilepsy with ragged red fibres (MERRF)	4	2014
SCARB2	Recessive	Action myoclonus renal failure (AMRF)	4	2014
KCNC1	Dominant	Myoclonus epilepsy and ataxia due to *KCNC1* (MEAK)	3	2021
DHDDS	Dominant	Developmental Epileptic Encephalopathy / PME	3	2021
GBA	Recessive	Gaucher disease	2	2014
CLN5	Recessive	Neuronal ceroid lipofuscinosis (NCL)	1	2014
CLN2	Recessive	Neuronal ceroid lipofuscinosis (NCL)	1	2014
NPC2	Recessive	Niemann-Pick disease, Type C	1	2014
HTT	Dominant	Huntington disease, Juvenile-onset	1	2014
ASAH1	Recessive	PME + Spinal muscular atrophy	1	2014
AFG3L2	Recessive	Spinocerebellar ataxia / PME	1	2021
NAXE	Recessive	Early onset encephalopathy / PME	1	2021
CHD2	Dominant	Developmental Epileptic Encephalopathy / PME	1	2021
CERS1	Recessive	PME due to impaired ceramide synthesis	1	2021
SACS	Recessive	Spastic ataxia / PME	1	2021
CACNA2D2	Recessive	Developmental Epileptic Encephalopathy / PME	1	2021
STUB1	Recessive	Spinocerebellar ataxia / PME	1	2021

*Four patients with Lafora disease were not submitted to genetic investigations but they were diagnosed by the finding of skin Lafora bodies and congruent clinical presentation

e Figure 1. Breakdown of cohort by both cluster analysis (2014) and clinical classification (2021) with indication of genetic status (solved versus unsolved).