Supplementary data

Figure S1 – Mutational and clinical items in MTM1-LOVD database, an entry example

A

Patient data (#0012336)	
Patient ID	[redacted]
Disease	myopathy, myotubular, type 1, X-linked (XL-MTM1)
Phenotype additional	Mild
Reference	Portugal:Porto
Remarks	-
# reported	1
Geographic origin	Portugal
Ethnic origin	-
Gender	F
Inheritance	isolated (sporadic)
Consanguinity	-
CK level	-
MR-IQ	normal
FVC	-
Age diagnosis	30y
Age onset	4-6y
Phenotype onset	Frequent falls
Motor_ability	Walks (only in flat surfaces)
Wheelchair_dependent	-
Age at death	>32y
Protein data	-
Remarks (non public)	Slowly progressive proximal weakness (especially in lower limbs); severe difficulty rising up from the floor and climbing stairs; moderate difficulty rising up from sitting. Positive Gowers sign.

B

Variant data	
Allele	Unknown
Reported pathogenicity	Pathogenic
Concluded pathogenicity	Unknown
Exon	12
DNA change	c.1262G>A (View in UCSC Genome Browser, Ensembl)
Var_pub_as	-
RNA change	r.(?)
Protein change	p.(Arg421Gln)
DB-ID	MTM1_00174
Variant remarks	conserved residue (C. elegans and/or S. cerevisiae R); functional effect (disruption of ligand binding site); de novo, in patient
PolyPhen	1.000
Genet_ori	de novo
Reference	-
Template	DNA
Technique	SEQ
Frequency	-
RE-site	-
DNA_g	-

Legend: Each database entry is subdivided into two tables: patient/clinical items (A) shared among the different LSDBs in the Leiden Muscular Dystrophy pages, and variant data (B).