LIST OF ABBREVIATIONS

4-MU	4-methyl umbeliferron
AF	amniotic fluid
ARMS-PCR	Amplification refractory mutation system - polymerase chain reaction
BSA	Bovine serum albumin
bp	Base pair
CV	Chorinic villus
CT	Computed tomography
CRF	Case record form
CNS	Central nervous system
cDNA	DNA complementary to RNA
DEB	DNA extraction buffer
DNA	Deoxiribonucleic acid
dNTPs	Deoxinucleotide triphosphate
EDTA	Ethylenediamine tetra-acetic acid
Etbr	Ethidium Bromide
ERNDIM	European Research Network for Evaluation and Improvement of Screening, Diagnosis, and Treatment of Inherited Disorders of Metabolism
GM1	N-acetyl-neuraminosyl-gangliotetraosylceramide
GM2	N-acetyl-neuraminosyl-gangliotriaosylceramide
GM3	N-acetyl-neuraminosyl-lactosylceramide
GlcNAc	N-acetyl galactosamine
GLSs	Glycosphingolipids
GM2A	GM2 activator protein
Hex-A	β-Hexosaminidase-A

Hex-B	β-Hexosaminidase-B
HAS	Human Serum Albumin
Kb	Kilo base pair
L	Liter
LSD	Lysosomal storage disorder
MRI	Magnetic resonance imaging
M	Mole
mM	Milimole
µM	Micromole
mg	miligram
ml	Milliliter
µl	Microliter
µg	Microgram
MLPA	Multiplex ligation dependent probe amplification
MW	Molecular Weight
MUGS	4-methylumbelliferyl-N-acetyl-β-D-glucosamine-6-sulphate
MUG	4-methylumbelliferyl-N-acetyl-β-D-glucosamine
nmol	Nanomole
nsSNP	Non-synonymous single nucleotide substitutions
PCR	Polymerase chain reaction (PCR)
PBS	Phosphate buffered saline
PD	Prenatal Diagnosis
Pmol	Picomole
RE	Restriction enzyme
RF	Relative fluorescence

RFLP	Restriction fragment length polymorphism
RMSD	Root mean square deviation
SD	Sandhoff disease
SDS	Sodium dodecyl sulphate
SIFT	Sorting intolerant from tolerant
TAE	Tris acetic EDTA
TE	Tris EDTA
Total-Hex	β-Hexosaminidase-total
TSD	Tay-Sachs disease
Taq	Thermus acquaticus
U	Unit
UV	Ultra violet