

Supplementary file 1. Summary of the study cohort

| Case study number | sex | Phenotype |
|-------------------|--------|-----------|
| 1 | Male | MEMSA |
| 2 | Male | MEMSA |
| 3 | Female | MEMSA |
| 4 | Male | MEMSA |
| 5 | Male | MEMSA |
| 6 | Male | MEMSA |
| 7 | Female | MEMSA |
| 8 | Female | MEMSA |
| 9 | Male | ANS |
| 10 | Male | MEMSA |
| 11 | Female | MEMSA |
| 12 | Female | MEMSA |
| 13 | Male | MEMSA |
| 14 | Female | MEMSA |
| 15 | Female | MEMSA |
| 16 | Male | MEMSA |
| 17 | Female | MEMSA |
| 18 | Female | MEMSA |
| 19 | Female | ANS |
| 20 | Female | MEMSA |
| 21 | Male | MEMSA |
| 22 | Female | MEMSA |
| 23 | Male | MEMSA |
| 24 | Male | MEMSA |
| 25 | Female | MEMSA |
| 26 | Female | MEMSA |
| 27 | Male | MEMSA |
| 28 | Female | Alpers |
| 29 | Female | ANS |
| 30 | Male | ANS |
| 31 | Female | ANS |
| 32 | Male | Alpers |
| 33 | Male | PEO |
| 34 | Male | Alpers |
| 35 | Male | Alpers |
| 36 | Male | Alpers |
| 37 | Male | Alpers |
| 38 | Female | Alpers |
| 39 | Female | MEMSA |
| 40 | Male | Alpers |
| 41 | Female | Alpers |
| 42 | Male | Alpers |
| 43 | Female | Alpers |

| | | |
|----|--------|--------|
| 44 | Female | Alpers |
| 45 | Female | Alpers |
| 46 | Female | Alpers |
| 47 | Female | MCHS |
| 48 | Male | MCHS |
| 49 | Male | MCHS |
| 50 | Female | MCHS |
| 51 | Male | Alpers |
| 52 | Male | Alpers |
| 53 | Female | Alpers |
| 54 | Male | MEMSA |
| 55 | Male | Alpers |
| 56 | Male | Alpers |
| 57 | Female | MEMSA |
| 58 | Female | Alpers |
| 59 | Female | MEMSA |
| 60 | Female | MEMSA |
| 61 | Male | Alpers |

ANS : Ataxia Neuropathy Sepectrum, MEMSA : Myoclonic Epil
MCHS: Myocerebrohepatopathy Spectrum

| Mutations (polymorphism) | Survival status | Anaemia |
|-----------------------------------|-----------------|---------|
| p.Ala467Thr/p.Ala467Thr | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Alive | NO |
| p.Ala467Thr/p.Trp748Ser | Deceased | Yes |
| p.Ala467Thr/p.Trp748Ser | Alive | NO |
| p.Ala467Thr/p.Trp748Ser | Deceased | NO |
| p.Ala467Thr/p.Trp748Ser | Deceased | Yes |
| p.Ala467Thr/p.Trp748Ser | Deceased | Yes |
| p.Ala467Thr/p.Trp748Ser | Deceased | Yes |
| p.Ala467Thr/p.Trp748Ser | Deceased | Yes |
| p.Ala467Thr/p.Trp748Ser | Alive | NO |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Alive | Yes |
| p.Trp748Ser/p.Trp748Ser | Alive | NO |
| p.Trp748Ser/p.Trp748Ser | Alive | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | NO |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Alive | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | NO |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Alive | NO |
| p.Trp748Ser/p.Trp748Ser | Alive | NO |
| Gly268Ala/Gly268Ala | Deceased | Yes |
| p.Ala467Thr/p.Gly303Arg | Deceased | Yes |
| p.Trp748Ser/p.Thr251Ile+Pro587Leu | Alive | NO |
| p.Ala467Thr/p.Gly303Arg | Deceased | Yes |
| p.Ala467Thr/p.Gly848Ser | Deceased | Yes |
| p.Ala467Thr/p.Gly303Arg | Deceased | Yes |
| p.Trp748Ser/ p.Gly1052Asp | Deceased | Yes |
| p.Trp748Ser/p.Arg807Cys | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Alive | Yes |
| p.Trp748Ser/p.Arg807Cys | Deceased | Yes |
| p.Ala467Thr/p.Gly303Arg | Deceased | Yes |
| p.Ala467Thr/p.Thr914Pro | Deceased | Yes |
| p.Ala467Thr/p.Gly848Ser | Deceased | Yes |

| | | |
|--|----------|-----|
| p.Arg1096Cys/p.Arg1096Cys | Unknown | NO |
| p.Arg807His/p.Gly1052Ser | Deceased | Yes |
| p.Ala467Thr/p.Ala467Thr | Alive | NO |
| p.His1110Tyr+ (p.Gln1236His) / p.Trp748Ser+(p.Glu1143Gly) | Deceased | Yes |
| p.Arg232His/p.Gly848Ser | Deceased | NO |
| p.Thr251Ile+p.Pro587Leu/p.Glu1136Lys | Deceased | Yes |
| p.Thr251Ile+p.Pro587Leu/p.Glu1136Lys | Deceased | Yes |
| p.Arg227Trp/p.Arg227Trp | Deceased | Yes |
| p.Gly848Ser/p.Lys498Thr | Alive | NO |
| p.Ala467Thr/p.Ala467Thr | Deceased | NO |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Trp748Ser | Deceased | Yes |
| p.Trp748Ser/p.Phe749Ser | Deceased | Yes |
| p.Ala467Thr/p.Trp748Ser | Deceased | NO |
| p.Trp748Ser/p.Gly848Ser | Unknown | NO |
| p.Ala467Thr/p.Trp748Ser | Deceased | NO |
| p.Ala467Thr/p.Ala467Thr | Alive | NO |
| p.Ala467Thr/p.Thr914Pro | Deceased | NO |

epsy Myopathy sensory ataxia, PEO: Progressive External Ophthalmoplegia ,