**Supplementary Table 1.** Clinical course of ATTR-FAP in family with Val71Ala TTR mutation

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| --- | --- | --- | --- | --- | --- |
| Patient no/ gender | Age at onset (yrs) | First manifestations | Clinical course | Ancillary studies | Survival and outcome |
| I generation | | | | | |
| 1 (I1) | mother of proband died of devastating disorder before 40 yr of life, similar symptoms were observed in 4 of her 5 children in next years | | | | |
| II generation | | | | | |
| 2 (II1 M  Proband) | 32 | pain and paresthesia in lower and upper limbs | severe sensory and motor polyneuropathy, impotence, diarrhea, constipation, cachexia | ENG:  sensory and motor  axonopathy,  sural nerve biopsy:  severe axonopathy and  amyloid deposits | 6 years,  died aged 38 |
| 3 (II7 M) | 38 | paresthesia  in lower and upper limbs | severe sensory and motor polyneuropathy, impotence, diarrhea, constipation, orthostatic hypotension  cachexia | ENG:  sensory and motor  axonopathy,  sural nerve biopsy:  severe axonopathy  amyloid deposits | 9 years  died aged 47 |
| 4 (II4 F) | 42 | paresthesia  in lower limbs | sensory and motor polyneuropathy, diarrhea, cachexia | ENG:  sensory and motor axonopathy | 6 years,  died aged 48 |
| 5 (II9 M) | 39 | NA | according to family history: progressive sensory symptoms and weakness, cachexia, before death blind and immobilized | NA | 5 years,  died aged 44 |
| III generation | | | | | |
| 6 (III6M, son of II4) | 44 | orthostatic hypotension | 46 yo gastrointestinal dismotility, paresthesia  48 yo sensory and motor polyneuropathy (walk with sticks)  cachexia | ENG:  sensory motor axonopathy,  sural nerve biopsy:  severe axonopathy  amyloid deposits | 4 years,  died aged 48 |
| 7 (III7M,  (son of II4) | NA | NA | according to family history immobilized and cachectic after few years of disorder | NA | died aged 49 |
| 8 (III15F, daughter of II9) | 39 | visual symptoms | 42 yo paresthesia in lower limbs, vitrectomy, 43 yo LT,  46 yo progression of polyneuropathy and cachexia | DNA analysis  TTR Val71Ala mutation | alive 51yr, walks with rollator, 12 years of disease, 8 years after LT |
| 9 (III17F, daughter of II9) | 29 | paresthesia in lower limbs | 32 yo visual symptoms – vitrectomy, 34 yo gastrointestinal dismotility, sensory and motor polyneuropathy  35 yo LT, 39 yo  wheelchair-bound  cachexia | ENG:  severe  sensory and motor axonopathy | 11 years,  died aged 40, 5 years after LT |
| IV generation | | | | | |
| 10 (IV1 M, son of III15) | 34 yo, Val71Ala TTR mutation carrier, no clinical signs and symptoms of TTR-FAP | | | | |

ENG – electroneurography; F – female; M – male; NA – not available; LT – liver transplantation