**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 MProband) | 32  | pain and paresthesia in lower and upper limbs | severe sensory and motor polyneuropathy, impotence, diarrhea, constipation, cachexia  | ENG: sensory and motoraxonopathy,sural nerve biopsy: severe axonopathy andamyloid deposits | 6 years,died aged 38 |
| 3 (II7 M) | 38  | paresthesiain lower and upper limbs | severe sensory and motor polyneuropathy, impotence, diarrhea, constipation, orthostatic hypotension cachexia | ENG: sensory and motoraxonopathy,sural nerve biopsy: severe axonopathyamyloid 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 axonopathyamyloid 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 polyneuropathy35 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