Parman\_Article 1\_Supplementary materials.

**Table S1.** European Expert (ATTReuNET) pre-meeting questionnaire 2012 and 2014

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| **Questions** |
| **1. The structure for care of rare diseases and TTR-FAP in your country** |
| Is there a Rare Disease Plan in your country?  |
| Are there identifiable expert/reference centres for the management of amyloid disease in your country? |
| Is there a specific professional society/group focused on amyloid-associated disease in your country? |
| Are there identifiable expert/reference centres specifically for the management of TTR‑FAP in your country? |
| Are there regional centres for the management of TTR‑FAP in your country? |
| Is there a specific TTR‑FAP patient support group in your country? |
| Approximately how many diagnosed TTR-FAP patients are there in your country? |
| Approximately how many people have been identified as carriers of a TTR gene mutation in your country? |
| Please give a broad picture of your patient cohort over the past 3 years |
| **2. Diagnosing TTR‑FAP** |
| For the diagnosis of TTR‑FAP, which types of biopsy are performed in your centre (in priority order with % use for each)? |
| What are the three most common genetic mutations associated with TTR-FAP in your patients and what percentage of your total patients do they represent? |
| How many mutations associated with TTR‑FAP have been identified in your country? |
| Do you routinely encourage genetic counselling to family members of diagnosed TTR-FAP patients? |
| Do you have access to specific expert genetic counsellors in your centre? |
| **3. Management of patients diagnosed with TTR-FAP and funding** |
| What is the mean waiting time for a TTR‑FAP patient before liver transplant?  |
| Is funding of care for TTR-FAP patients an issue in your country? |
| Do people with a diagnosis of TTR-FAP receive specific social benefits/social security in your country? |
| **4. On-going care of TTR-FAP patients and asymptomatic carriers** |
| For how long do you typically manage a TTR‑FAP patient in your centre? |
| How frequently does a TTR-FAP patient visit your clinic for routine follow up?  |
| How many specialists will a TTR-FAP patient see at each follow-up visit? |
| Which investigations and tests do you undertake at a routine review of TTR-FAP patients?  |
| Which investigations and tests do you undertake at a routine review of asymptomatic carriers? |
| How frequently does an identified asymptomatic carrier of a TTR mutation visit your clinic for routine follow up?  |
| **5. The typical ‘patient journey’ in your country** |
| In your experience, how many different physicians does the typical TTR-FAP patient see prior to achieving an accurate diagnosis? |
| In your experience, what would you estimate is the average amount of time between symptom onset and accurate diagnosis of TTR-FAP?  |
| In your experience, what is the typical ambulatory status of patients at diagnosis of TTR-FAP (stage and proportion %)?  |
| **6. Further considerations** |
| Which factors in the current management of TTR-FAP in your country do you think work well? |
| Which factors in the current management of TTR-FAP in your country do you think could be improved? |
| What do you feel are the barriers to optimal diagnosis and care of TTR-FAP patients in your country? |

TTR, transthyretin; TTR-FAP, transthyretin familial amyloid polyneuropathy.

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| TABLE S2. Typical patient journey in European countries |  |
|  | **How many different physicians does the typical TTR-FAP patient see prior to achieving an accurate diagnosis?** | **Average time to diagnosis since first symptoms** | **Multidisciplinary approach****(number of specialists seen at each follow-up)** |
| **Portugal** | 1–2 | 2 years with family history3–5 years without | > 2 |
| **Sweden** | 1–2 | < 2 years in endemic area2–5 years in non-endemic area | 1–2 |
| **Cyprus** | 1–2 | < 2 years | > 2 |
| **France** | 3–4 | 1–3 years with family history2–4 years without | 1–2 |
| **Italy** | 3–4 | 2–3 years | > 2 |
| **Spain**  | 3–4 | 1–4 years | > 2 |
| **Bulgaria** | 3–4 | 3 years | > 2 |
| **Germany** | 3–4 | ~ 3 years | > 2 |
| **Netherlands** | 3–4 | < 2 years | 1–2 |
| **Turkey** | 3–4 | 3–5 years | > 2 |

TTR-FAP, transthyretin familial amyloid polyneuropathy.

Information compiled from clinical experience of the European Network for TTR-FAP (ATTReuNET) in March 2014.

**Table S3.** Expert participants of the European Network for TTR-FAP (ATTReuNET) 2012 and 2014

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| --- | --- | --- | --- | --- | --- | --- |
| **Expert** | **Speciality** | **Country** | **Town****centre** | **National Reference Centre** | **For FAP or amyloidosis** | **National network**  |
| Teresa Coelho | Neurology | Portugal | Porto | Yes | FAP | - |
| Ole B. Suhr | Internal medicine,gastroenterology | Sweden | Umeå | Yes | FAP | - |
| David Adams | Neurology | France | Paris | Yes | FAP | +  |
| Michel S. Slama | Cardiology | France | Clamart | Yes | FAP | + |
| Laura Obici | Internal medicine  | Italy | Pavia | Yes | Amyloidosis | +  |
| Ernst Hund | Neurology | Germany | Heidelberg | Yes | Amyloidosis | +  |
| Bouke P. Hazenberg | Internal medicine, rheumatology | The Netherlands | Groningen | Yes | Amyloidosis | + |
| Jan B. Kuks | Neurology | The Netherlands | Groningen | Yes | Amyloidosis | + |
| Juan Buades  | Internal medicine | Spain | Palma de Mallorca | No | FAP | - |
| Josep M. Campistol | Nephrology | Spain | Barcelona | No | Amyloidosis | - |
| Lucia Galan | Neurology | Spain | Madrid | No | FAP |  |
| Theodore Kyriakides | Neurology | Cyprus | Nicosia | Yes | FAP | - |
| Yesim Parman | Neurology | Turkey | Istanbul | Yes | FAP | + |
| Ivailo Tournev | Neurology | Bulgaria | Sofia | Yes | FAP | - |
| Velina Guergueltcheva | Neurology | Bulgaria | Sofia | Yes | FAP | - |

ATTReuNET, European Network for TTR-FAP; FAP, familial amyloid polyneuropathy; TTR-FAP, transthyretin familial amyloid polyneuropathy