TTR-FAP Italian Registry: a collaborative network for definition of natural history, psychosocial burden, standards of care and clinical trials

Transthyretin-related familial amyloid polyneuropathy (TTR-FAP) is an inherited severe and disabling disease causing sensory and motor neuropathy, autonomic disabilities, and cardiomyopathy. Liver transplantation is currently the only treatment for preventing toxic amyloid deposition in the organs. Since 2011, tafamidis meglumine is available in Italy only for ambulant patients without any help and it seems promising, but now new disease-modifying RNA therapies are on the horizon. There are still many unresolved questions in the genotype-phenotype correlation and about inter- and intra-mutation variability. Moreover, there are very few data on longitudinal functional changes, psychosocial burden of patients and caregivers and epidemiological distribution in Italy. We are developing a TTR-FAP National Registry, where we will collect clinical and genetic information of the disease, and measure disease course in 1-year period. We plan to follow the patients by several already validated scales and tools to investigate neurological, cardiological and psychological impairment. 6-minute walk test will be validated as a reliable measure for future trials. Participants in the National Registry will be asked to give a written informed consent and to choose the nearest clinical centre among nine centres, where he/she wants to be followed during the 1-year longitudinal study. We expect to enroll approx. 272 symptomatic patients and approx. 100 asymptomatic TTR gene mutation carriers. Data will be then analysed and will provide epidemiological data on Italian TTR-FAP patients, define natural history of the disease, standards of care, disease burden and patients' needs, and facilitate feasibility and planning of future clinical trials