

Progetti innovativi per le malattie rare

Patient-Reported Outcome Measures nell'Amiloidosi Cardiaca da Transtiretina Wild-Type e Variante: lo Studio ITALY

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Abstract

Specific, standardized, comprehensive, universally accepted Patient-Reported Outcome Measures (PROMs) are currently lacking for variant and wild-type cardiac transthyretin amyloidosis (v-ATTR/wt-ATTR). We aim to create two scores providing a cumulative assessment of cardiac involvement, peripheral neuropathy and comorbidities, and their impact on the quality of life.

In the setting of a nationwide collaboration involving 5 main Italian referral centers (in Bologna, Firenze, Pavia, Pisa and Messina), a panel will be created, including experts of ATTR cardiomyopathy, neurologists, geriatricians, health management specialists, as well as patients with either v-ATTR or wt-ATTR cardiomyopathy.

The most clinically relevant domains for patients (such as physical limitations, symptoms, self-efficacy and knowledge, social interference, quality of life, age-related issues, social and family environment, frailty, comorbidities) will be identified. Two sets of 25 items (one for variant and another for wild-type ATTR cardiomyopathy) will be created in collaboration with patients. Questions will be formatted for gender neutrality, clarity, interpretability, and possible foreign language translations. PROMs scores will be validated through administration to around 250 consecutive outpatients. Score performance will be evaluated in terms of internal consistency, response to clinical changes, comparison with conventional clinical measures. The time needed for completion, the clarity of questions and the need for assistance from a family caregiver will be evaluated.

This project will hopefully lead to the identification of disease-specific metrics that may serve as a clinically meaningful outcome in cardiovascular research, patient management, and quality assessment.