Design and initiation of the Croatian Transthyretin Cardiac Amyloidosis Registry

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Introduction: Transthyretin amyloidosis (ATTR) is a rare disease with heterogeneous symptoms and unfavorable outcomes unless diagnosed and treated in the early stage. Phenotypes and clinical presentations relate to underlying genetic variants (where genotype heterogeneity is well-known and related to endemic geographic regions) or the acquired form (wild type)[1,2]. The Croatian Transthyretin Cardiac Amyloidosis (CroATTR) Registry is designed as a national, longitudinal, non-interventional, and both retrospective and prospective ATTR registry.

Methods: We aim to include patients with clinically proven hATTR-CM or wtATTR-CM according to the current guidelines, or family members with confirmed mutation of the TTR gene (regardless of the presence of cardiomyopathy). The registry will acquire basic demographic characteristics and results of genetic testing (for hATTR), followed by clinical work-up capturing patient demographics, quality of life questionnaires, medical and family history, data from 12-lead electrocardiogram (ECG), echocardiography, cardiac magnetic resonance imaging (cMRI) (with an emphasis on typical ATTR red flags), 99mTc- pyrophosphate scintigraphy, electromyoneurography, and myocardial biopsy, as available. The registry will follow disease-specific outcomes: 1. overall survival, 2. cardiovascular mortality, 3. heart failure hospitalizations/unscheduled physician visits, 4. patient reported outcomes in the area of quality-of-life changes. The registry will also collect data on disease- specific treatments in our population: the proportion of patients treated with of guideline directed medical therapies (GDMT) for amyloidosis and heart and/or liver transplantation. The data will be captured at the time of inclusion of the patient in the registry (including retrospective data focusing on the time the diagnosis was first made) and will include prospective recurring visits. Data will be collected and managed using REDCap electronic data capture tools (the design of the database is shown on **Figure 1**).

Conclusion: The CroATTR Registry will aggregate ATTR patients and allow further insights into the occurrence and natural course of disease. A particular emphasis will be made on the rare genetic mutation prevalent in our population, the utilization of guideline directed medical therapies and transplantation procedures.

Data Collection Instrument	Data at registry initiation	Retrospective data	Recurring visit	Final visit	Outcomes
Baseline patient information	●				
Inclusion criteria	●				
Medical history	●				
Family History	●				
Vital signs and antropometric data	●	●	●	●	
Physical status and functional class	●	●	●	●	
Nuclear scintigraphy	●				
Tissue biopsy	●				
ECG	●	●	●	●	
Echocardiographic data	●	●	●	●	
Cardiac Magnetic Resonace Imaging	●	●		●	
Electromyoneurography	●	●	●	●	
Laboratory findings	●	●	●	●	
Medication and therapies	●	●	●	●	
Specific TTR therapy	●	●	●	●	
KCCQ Form	●		●	●	
EQ5D	●		●	●	
Disease outcomes			●	●	●

FIGURE 1. Croatian Transthyretin Cardiac Amyloidosis Registry Electronic Case Report Form.

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LITERATURE

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