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Expanding the Genotypic Spectrum of Hereditary Transthyretin Amyloidosis: A Case Series of Patients with the Novel TTR p.Ala101Gly Variant

Hereditary transthyretin amyloidosis (ATTRv) is a rare autosomal dominant multisystem disorder caused by pathogenic variants in the transthyretin (TTR) gene, resulting in progressive amyloid deposition in peripheral nerves and the myocardium. The rare TTR c.302C>T (p.Ala101Val) variant has been reported predominantly in patients with cardiac ATTRv, but clinical data remain limited. The aim of this study was to characterize the clinical phenotype associated with this variant in a Belarusian cohort. We analyzed clinical and genetic data from five unrelated Belarusian probands with ATTRv treated between 2024 and 2025. The pathogenic TTR c.302C>T (p.Ala101Val) variant was identified by Sanger sequencing in all patients. Diagnosis was confirmed by exclusion of AL amyloidosis and positive technetium-99m bone scintigraphy, with histopathological confirmation available in four cases. All patients demonstrated late disease onset (64-74 years). Four probands presented with a predominant cardiac phenotype characterized by amyloid cardiomyopathy, ventricular hypertrophy, heart failure, conduction disturbances, and atrial or ventricular arrhythmias, whereas one patient manifested primarily with recurrent arrhythmic events without overt cardiomyopathy. Neurological involvement was identified in all patients. Two individuals developed advanced biventricular heart failure and died within one year of diagnosis. Cardiac magnetic resonance imaging demonstrated extensive myocardial infiltration, while bone scintigraphy showed only low-to-moderate myocardial tracer uptake (Grade 1-2) despite significant cardiac involvement. Pedigree analysis confirmed autosomal dominant inheritance with incomplete, likely age-dependent penetrance. In conclusion, the TTR p.Ala101Val variant is associated with a late-onset ATTRv phenotype dominated by cardiomyopathy, arrhythmias, and neurological involvement. The relatively low sensitivity of bone scintigraphy in these patients highlights the importance of genetic testing and genotype-informed diagnostic strategies in suspected ATTR cardiomyopathy.

Keywords: hereditary transthyretin amyloidosis, amyloid cardiomyopathy, polyneuropathy, genetic analysis, endemic mutation

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Biography

Nadiia M. Rineiska, MD, PhD, is a researcher at the Laboratory of Chronic Heart Failure, State Institution Republican Scientific and Practical Centre "Cardiology" (Minsk, Belarus). Her research focuses on hypertrophic cardiomyopathy, arrhythmogenic cardiomyopathy, cardiac amyloidosis, Fabry disease, and other inherited and infiltrative cardiomyopathies. Her work encompasses molecular genetics, precision diagnostics, risk stratification, and prevention of adverse outcomes in patients with rare cardiovascular diseases. Dr. Rineiska is actively involved in clinical and translational cardiovascular research and has authored numerous publications in the fields of cardiomyopathy and heart failure.

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