Conference Proceedings By
United Research Forum, London, UK

ORGANIZED BY
UNITED RESEARCH FORUM
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World Congress on

Dementia, Alzheimer's, Parkinson's & Neurodegenerative Disorders

July 09-10, 2026 | Frankfurt, Germany

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Hereditary transthyretin-related amyloidosis is frequent in polyneuropathy and cardiomyopathy of no obvious aetiology

Hereditary Transthyretin-Related Amyloidosis (hATTR), a clinically heterogeneous autosomal dominant disease caused by pathogenic variants in the TTR gene. The study objective was to define the prevalence of hATTR in patients with polyneuropathy and/or cardiomyopathy of no obvious aetiology.

A multicenter observational "Epidemiological analysis for the hereditary Transthyretin-Related AMyloidosis"-TRAM study was performed in Germany, Austria, and Switzerland.

doi.org/10.51219/URForum.2026.Volha-Skrahina

ISBN: 978-1-917892-37-7

A total of 5141 participants were recruited by 50 neurologic and 27 cardiologic specialized centres. Genetic analysis demonstrated a 1.1% hATTR positivity rate. Twenty-one various TTR variants were identified. Body Mass Index was lower in the TTR-positive patients as an indicator for the involvement of the autonomic nervous system; the age of onset of clinical manifestations was higher in TTR-positive patients. There were no other genotype-phenotype correlations or the prevalence of specific clinical manifestations in TTR-positive patients.

Our data support the fact that Hereditary Transthyretin-Related Amyloidosis is clearly underdiagnosed in polyneuropathy and cardiomyopathy patients. Routine implementation of genetic testing is important and crucial in patients with unexplained polyneuropathy and/or cardiomyopathy to accelerate the earlier diagnosis and the time-sensitive treatment initiation.

Keywords: Hereditary transthyretin-related amyloidosis; cardiomyopathy; genetic testing; polyneuropathy.

Biography:

Volha investigated the translational regulation of the innate immune response in tuberculosis infection during her Master's thesis at the Max Planck Institute in Berlin, Germany. She has completed her PhD thesis on the host fungal micronutrient interaction at the Hans Knoell Institute in Jena, Germany in 2018. Volha has worked as a Director of Clinical Studies at CENTOGENE and as a Scientific Manager in Rhythm Pharmaceuticals. She executed multiple international clinical studies to analyse the prevalence of rare diseases allowing patients to be diagnosed and obtain treatment at the earliest possible moment.

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