ATTRv: a Multisystemic Disease Requires a Multiprofessional Approach

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Pathogenic variants of the transtiretin gene (TTRv) result in the production of amyloidogenic fibrils that deposit in the extracellular space of many tissues, resulting in amyloidogenic multisystemic disease (ATTRv). Several tissues may be affected in different combinations, including the peripheral nerves and the cardiac system, which are usually the main targets, but also the kidneys, eyes, gastrointestinal system, central nervous system and musculoskeletal structures.¹

ATTRv is a progressive disease² that results in death around 8 to 15 years after the onset of the disease, even more rapidly when the cardiac system is involved early.³ However, the natural history of the disease has changed with the introduction of treatments that, by different mechanisms, decrease the production of amyloid fibrils.⁴

Such a complex disease demands coordinated multiprofessional interaction in order to provide the best

care for patients. Considering medical specialties, well trained cardiologists, neurologists, ophthalmologists, gastroenterologists, nephrologists and geneticists should work in close collaboration both to treat patients who have already manifested the disease and to follow asymptomatic carriers, in order to detect the disease onset, when treatment is much more effective. For the best care, however, in addition to professionals who are trained in the field of amyloidosis, it is necessary that an institution is able to provide a complex set of prepared investigations, including neurophysiology (including small fiber evaluation), skin biopsy, other types of biopsy, magnetic resonance image, and scintigraphy.

For these reasons, we believe these patients should be followed in tertiary reference centers that should interact closely with local physicians, in order to offer the best care at the best time to patients with this complex disease.

Keywords

Cardiomyopathy; Polyneuropathy; Transthyretin; TTR.

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Manuscript received September 19, 2021, revised manuscript September 26, 2021, accepted September 26, 2021.

DOI: <https://doi.org/10.36660/abchf.20210026>

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