Role of Nerve Conduction Study in monitoring ATTRv patients

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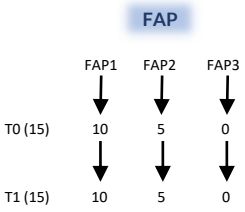
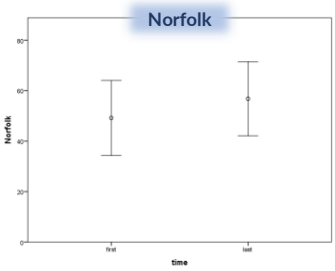
Background

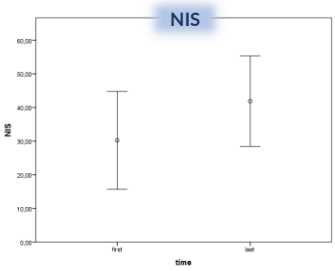
Hereditary TTR Amyloidosis (ATTRv) is a rare disease characterized by progressive neuropathy. Patisiran is an RNA interference agent that inhibits the hepatic synthesis of transthyretin. Data from real-world studies have confirmed the role of this therapeutic agent in halting disease progression, although long-term follow-up has shown an increased disability burden, as assessed by the Neuropathy Impairment Score (NIS). The aim of this work was to explore the role of nerve conduction studies (NCS) in identifying changes during long-term follow-up.

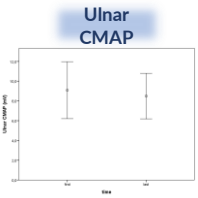
Methods

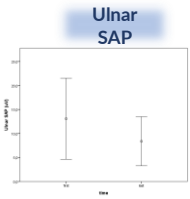
We enrolled genetically confirmed ATTRv patients with peripheral neuropathy undergoing therapy with Patisiran. Clinical and electrophysiological data were collected from the baseline visit and the most recent follow-up. Clinical data, including FAP stage, NIS, and the Norfolk quality of life questionnaire were collected. Additionally, electrophysiological data included the amplitude [distal Compound Muscle Action Potential (dCMAP), Sensory Action Potential (SAP)] and velocity of the peroneal, posterior tibial, sural, motor and sensory ulnar nerves. Statistical analysis was performed using paired t-tests to compare continuous variables and chi-squared tests for categorical variables between baseline and follow-up. Differences were considered statistically significant if the p-value was < 0.05.

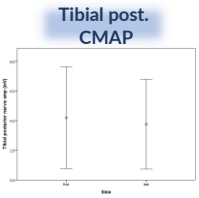


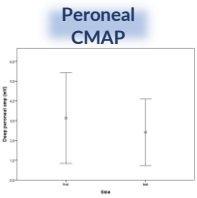


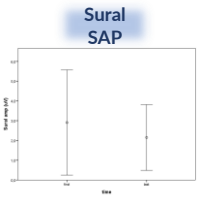












Results

Fifteen symptomatic patients were recruited [M/F = 10/5; age at first evaluation = 58.5±16.9 (31-80)]. Three main TTR pathogenic mutations were present in our cohort (Val30Met 8/15; Phe64Leu 4/15; Glu54Lys 3/15). At the baseline visit, 10 patients were in FAP1 stage and 5 in FAP2 stage. The mean follow-up duration was 28.2±13.5 months (12.2–51.8). During the follow-up period, **all patients remained in the same FAP stage category**, while **NIS significantly increased** (30.2±26.3 vs. 41.8±24.2; **p= 0.022**). However, **electrophysiological parameters remained unchanged** at the follow-up visit (dCMAP ulnar= 9±5.2 vs 8.4±4.1, p= 0.4; dCMAP peroneal= 3.1±4.1 vs 2.4±3, p= 0.1; dCMAP posterior tibial= 4.2±5.9 vs 3.7±5.2, p= 0.4; SAP ulnar= 13±15.2 vs 8.3±9.1, p= 0.05; SAP sural= 2.9±4.6 vs 2.1±2.8, p= 0.3).

Conclusion

This long-term clinical and electrophysiological follow-up of ATTRv patients treated with Patisiran demonstrated that, despite the worsening of the NIS, no changes were observed in clinical staging (FAP), quality of life or electrophysiological parameters.

The study highlights **the limitations of NIS and emphasizes the important role of NCS** in monitoring disease progression and therapeutic responses in ATTRv patients.

24-28 Ottobre 2025
Padova Congress
55° CONGRESSO SOCIETÀ ITALIANA DI NEUROLOGIA