PNS 2023 Annual Meeting

Abstract Supplement

Copenhagen, Denmark

Hereditary transthyretin amyloidosis (hATTR) with polyneuropathy in North Macedonia: current evidence and experiences

Poster No:
P 009

Authors:
Ivan Barbov[1], Goce Kalcev[2]

Institutions:
[1]University Clinic of Neurology, 1000 Skopje, Republic of North Macedonia, Skopje, North Macedonia, [2]National Alliance for Neuromuscular Diseases and Neuroscience, GANGLION, Republic of North Macedonia, Skopje, North Macedonia

Introduction:
Hereditary transthyretin amyloidosis (hATTR) with polyneuropathy is a rare disease due to mutations in the gene encoding transthyretin (TTR), identified by multisystem extracellular deposition of amyloid and leading to dysfunction of different organs and tissues. hATTR amyloidosis represents a diagnostic challenge for neurologists considering the wide variability in clinical presentation and multiorgan involvement.

Methods:
To highlight the recent information regarding hATTR patients in North Macedonia.

Results:
In our country, hATTR was found in 23 patients between 2017 and 2022. 1 patient has the Val33Phe mutation, while 22 patients have the Glu89Gln mutation. 12 patients are currently receiving treatment with Tafamidis, while 4 patients are on the waiting list to begin therapy for the second stage of polyneuropathy, considering the nation's Rear Disease Committee. During this time, 4 patients passed away: 2 in the second stage of polyneuropathy, 1 in the third stage of polyneuropathy, and 1 with primarily cardiac symptoms. Currently, there are 18 asymptomatic carriers who are related to symptomatic hATTR patients. According to the geographic distribution, the majority of patients originate from the eastern part of the country.

Conclusions:
hATTR amyloidosis is a severe, clinically and genetically heterogeneous, multisystem disease that affects people all over the world. In order to accelerate the earlier diagnosis and the time-sensitive treatment start, routine genetic testing is advised for patients with unexplained polyneuropathy.

References:
No

References 1:

References 2:

References 3:

References 4:

Grant Support: -

Keywords: amyloid, polyneuropathy, hereditary, therapy, transthyretin