Neurological aspects in a case of amyloidosis transthyretin type variant Glu54Gln

D. Mirela[1,2,3,4,5], M. Draghici[5]
[1]*Neurology, Valciu Crisanda, Romania*
[2]*Hematology, Andreea Jercan, Romania*
[3]*Hematology, Daniel Coriu, Romania*
[4]*Hematology, Badelita Sorina Nicoleta, Romania*
[5]*Hematology, Daniel Coriu, Romania*

Transthyretin hereditary amyloidosis is a rare disease and, so far, this has considered limited to certain geographical areas. Recently, with the intensification of genetic screening possibilities, new cases, new mutation and endemic areas (e.g. the Balkan area in Europe) have been found-this shows that this disease is more widespread than was believed, so doctors need to be warned and think more often about this disease. We present the case of a 51 year old male patient, that had a sister who died at the age of 49 from a cardiac disease and his mother died at the age of 50. The onset of his disease was in 2012 with diarrhea of unknown cause (rectum biopsy – Congo Red negative). Afterwards, in 2014, he presented paresthesias and muscle weakness in the lower limbs. In March 2016 he was admitted to the Neurology Clinic-Fundeni and the EMG established the diagnosis of sensorimotor axonal polyneuropathy. The patient was then admitted to the Hematology Clinic with peripheral edema, painful lower limb paresthesias that progressed to knee-level, orthostatic hypotension, bowel disorders and cachexia. It was established to be Familial amyloidosis transthyretin type (Glu54Gln) with systemic involvement: Cardiac (restrictive cardiomyopathy), SNP (Sensorimotor axonal polyneuropathy), SNV(orthostatic hypotension). Familial amyloidosis transthyretine type is an incurable disease, with a median survival of 7-10 years after the onset of symptoms. Liver transplantation is the standard therapy for eligible patients because it stops the synthesis of the mutant TTR. Tafimidis and other transthyretin stabilizers are new agents, still under investigation.