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## Genotypic and phenotypic presentation of TTR-FAP in Turkey

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**Introduction:** Transthyretin-related familial amyloid polyneuropathy (TTR-FAP) is an autosomal dominant disorder caused by mutations of the transthyretin (TTR) gene. More than 100 different mutations of the transthyretin gene were identified worldwide, but still the first described Val30Met is the most common one. The mutant amyloidogenic transthyretin protein causes systemic accumulation of amyloid fibrils that results in organ dysfunction and death. TTR-associated FAP is a progressive and fatal disease if left untreated and should be considered in the differential diagnosis of any patient with a progressive polyneuropathy, especially with an accompanying autonomic involvement.

**Methods:** We studied clinical, electrophysiological, histopathological, and genetic characteristics in 14 Turkish patients (4 female, 10 male) from 9 families with polyneuropathy and mutations in TTR.

**Results:** Mean age of onset was  $43.6 \pm 13.3$  years (between 21-66 years). 9 of them were late-onset TTR-FAP. At onset, all the patients exhibited sensory loss of the lower and upper limbs, three patients also experienced severe autonomic symptoms. 5 patients had autonomic nervous system manifestations, and nine demonstrated evidence of amyloid cardiomyopathy, 2 of them had renal involvement. 5 patients (4 male) had carpal tunnel syndrome. 1 patient with Gly53Glu mutation showed episodes of dysarthria and hemiparesis which were already described to be associated with this genotype. 4 patients died during follow-up due to the systemic involvement. Sequence analysis of TTR gene revealed the presence of 6 different mutations (Val30Met [in 3 unrelated families], Glu89Gln, Gly53Glu, Glu74Gly, Gly47Glu, Glu109Gly).

**Conclusions:** Our study suggests that the TTR-FAP patients from Turkey exhibit a wide clinic and genetic heterogeneity.

**Disclosure:** Nothing to disclose