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We report a case of a rare TTR mutation in a patient with familial amyloidotic polyneuropathy (FAP).
Case presentation:
A 53-y old male presented in spring 2009 with a complex symptomatology of back pain, weakness and myalgia after heavy exercise. Work-up revealed a discal hernia with myelopathy as well as a slight polyneuropathy of unknown origin.

The patient father had died with coronary heart disease at the age of 48, and suffered from a neurologic disorder, first thought to be Guillain-Barré. The medical chart, dating from the 80's, was unavailable.
The patient underwent neurosurgery, unfortunately, the suggested synchrone nerve biopsy was not realised at that moment.

Extensive blood analyses were normal, including light chain assay and immunofixation. Serology of borrelosis was positive, also with western blot and associated with proteinorachia.

Post-operatively, the patient noted prolonged sustained fatigue as well as polyneuropathic symptom worsening. Worsening of the known axonal sensory deficit with proximal extension of the upper and lower limbs was confirmed by EMG. A trial of Ceftriaxone brought no benefit, and a trial of polyclonal immunoglobulin infusions was likewise unsuccessful. Finally, sural nerve biopsy was performed in December 2010, revealing amyloid deposits with moderate demyelination; there was neurogenic muscle degeneration. Research of the most common mutation of the TTR gene (ValMet30) was negative. DNA sequencing revealed the rare p.D59V mutation of the TTR gene. The patient finally retrieved medical chart of his father, showing that he also probably suffered from TTR amyloidosis.

Conclusion: This is the first report of a p.D59V mutation of the TTR gene in a Belgian patient. To our knowledge, one similar case has been documented in Germany. The patient underwent successful liver transplant in September 2011.