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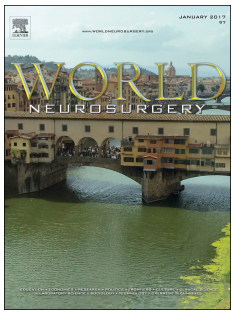

Oculoleptomeningeal amyloidosis secondary to the rare Transthyretin c.381T>G (p.Ile127Met) mutation

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Oculoleptomeningeal amyloidosis secondary to the rare Transthyretin c.381T>G (p.Ile127Met) mutation

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Key Words
Familial Amyloidosis, Oculoleptomeningeal Amyloidosis, Transthyretin, TTR

Abbreviations List
CNS – central nervous system
CSF – cerebrospinal fluid
MRI – magnetic resonance imaging
OLMA – oculoleptomeningeal amyloidosis
OMIM – Online Mendelian Inheritance in Man (genetic compendium database)
TTR - transthyretin

Abstract

Background: Oculoleptomeningeal amyloidosis (OLMA) represents a rare subtype of familial transthyretin (*TTR*) amyloidosis, characterized by deposition of amyloid in cranial and spinal leptomeninges along with ocular involvement. Of more than 100 *TTR* mutations identified, few have been associated with OLMA. Herein we describe the first report of leptomeningeal amyloidosis associated with the c.381T>G (p.Ile127Met) TTR mutation, linking this variant in the OLMA phenotype. ***Case Description:*** A 53 year-old male presented with a 2-year history of progressive symptoms including upper and lower limb weakness, ataxia, peripheral and autonomic neuropathy. Neuroimaging including gadolinium-enhanced MRI of the brain and spinal axis identified diffuse leptomeningeal enhancement along the brainstem and spinal cord plus evidence of hemosiderosis. Pathologic and genetic analyses of biopsy material from enhancing intradural extramedullary tissue at the thoracolumbar junction was diagnostic of amyloidosis of a transthyretin type secondary to a TTR c.381T>G (p.Ile127Met) mutation. ***Conclusions:*** OLMA represents a rare subtype of heritable transthyretin amyloidosis that may present with progressive neurologic decline secondary to central nervous system leptomeningeal amyloid deposition. This case identifies the c.381T>G (p.Ile127Met) *TTR* mutation variant as being implicated in the OLMA phenotype.