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Title	Oculoleptomeningeal Amyloid Val30Gly TTR Mutation
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Aim	Hereditary amyloidosis is a group of autosomal dominant diseases characterized by extracellular deposition of amyloid in various target organs, and classified biochemically by the implicated precursor protein, the most common being transthyretin. Transthyretin amyloidosis has over 100 genetic variants, but peripheral neuropathy (familial amyloidotic polyneuropathy) is the most common clinical manifestation. A much less common form manifests with CNS signs and symptoms and visual disturbances, aka occuloleptomeningeal amyloidosis. These patients often develop seizures, dementia, stroke-like episodes, and ataxia, and require vitrectomy for vitreous amyloid opacities. Several kindreds with these clinical and pathologic features have been described, one of which is characterized by a Val30Gly *TTR* mutation, the result of a point missense mutation in the transthyretin gene.
Materials & Methods	We describe the clinicopathologic features of one patient whose father had Val30Gly transthyretin amyloidosis.
Result	The patient was well until age 56 years when she presented with severe headaches and later with focal sensory deficits, concerning for stroke. Because of her family history, she refused TPA (tissue plasminogen activator) therapy. Several years later she developed decreased visual acuity and required bilateral vitrectomies. The diagnosis of amyloidosis was confirmed on vitrectomy specimen. In the subsequent years, she experienced intermittent episodes of loss of consciousness and problems with speech. She ultimately succumbed to metastatic colon cancer. Autopsy examination showed thickened, slightly pink, irregular leptomeninges over the base of the brain and surrounding the spinal cord. Microscopically, amyloid was deposited in the leptomeningeal vessel walls, subarachnoid space and choroid plexus. No infarcts, plaques or tangles were identified. General autopsy did not reveal amyloid elsewhere in the body.
Conclusion	Occuloleptomeningeal amyloidosis is exceedingly rare, but awareness of this entity is very important given the clinical overlap with other more common entities and because of the genetic implications of the diagnosis.