Vitreous amyloidosis is a rare condition that occurs in some forms of Transthyretin hereditary Amyloidosis, mainly in Familial Amyloidotic Polyneuropathy type I. Vitreous opacities may be the earliest occurring or, in some cases, only symptom of this disorder. In such cases a family history of amyloidosis is usually present. We report on a case of vitreous amyloidosis in an 80-year-old man where there is no evidence of systemic involvement and no family history of amyloidosis.

Abbreviations: FAP – Familial Amyloidotic Polyneuropathy, TTR – Transthyretin

Introduction

Amyloidosis is the term used for a group of disorders, of diverse etiology, characterized by the extracellular accumulation of insoluble fibrillar substances, collectively called ‘amyloid’, in various organs and tissues. Any one of a variety of proteins may cause amyloidosis, including immunoglobulin light chains, AA protein, beta-2-microglobulin, prion protein, transthyretin (TTR), beta-amyloid precursor protein, amylin, atrial natriuretic factor, procalcitonin, gelsolin and cystatin-C [1].

Vitreous deposits of amyloid are frequently found in some forms of Transthyretin hereditary amyloidosis, mainly in Familial Amyloidotic Polyneuropathy (FAP) type I, in which peripheral polyneuropathy with sensory and motor disturbances, autonomic dysfunction and cardiomyopathy are the prominent indications of the disorder [2]. Occasionally, however, the vitreous opacities are the earlier occurring and, sometimes, sole symptom of FAP. In such cases a family history of amyloidosis usually exist [3].

We report a case of vitreous amyloidosis with no evidence of systemic involvement and where the patient had no family history of amyloidosis.

Case report

The 80-year-old male patient was first examined in October 1991 for decreasing visual acuity, bilaterally, over the previous 3 months. His medical history revealed nephrectomy for renal tuberculosis in 1966 and the inset of Parkinson disease over the previous years. Visual acuity was finger counting at one meter R.E. and 20/80 L.E. Minor senile cataracts were found in both eyes. The vitreous body of his right eye was full of yellowish-white opacities (see Fig. 1) precluding examination of the fundus. Thin veil-like opacities were observed in his left eye but the retina appeared normal. Ocular movements and pupillary reflex were also normal. The intraocular pressure by applanation was 18 mmHg in both eyes. A general physical examination turned out normal. There were no neurological deficits or macroglosia nor was there
enlargement of the heart, liver or spleen. Serum and urine protein electrophoresis, electrocardiogram and electromyogram were also normal. Biopsy specimen of rectum did not reveal amyloid on a Congo Red stain test.

Investigation into his family history did not turn up evidence of familial amyloidosis in any relative. His mother died at age 28 years from influenza and his father was in good health until he died suddenly from an unknown cause at age 60 years and was reported to have had good sight. The patient’s sister, age 74 years, was living in another region and refused ophthalmologic examination but she was known to be in good health and to have good sight. We also found no ocular abnormalities in the son of the patient.

Although there was neither a family history of nor did the patient himself have polyneuropathy, autonomic neuropathy or gastrointestinal disease, amyloidosis of the vitreous body was suspected by ophthalmologic examination.

In January 1992 the patient underwent a pars plana vitrectomy on his right eye. Examination of a Congo Red-stained specimen of the vitreous under polarized light showed yellowish green birefringence consistent with amyloid. Immunohistochemical staining was positive for transthyretin. The blood lymphocitic DNA was examined using PCR amplification and restriction endonuclease digestion and the mutation TTR-Met-30 was found in the patient but not in his son.

Postoperatively his visual acuity corrected to 20/60 O.D. and small perivascular deposits on his arterioles and venules appeared.

Two months later the vitreous opacities of the left eye became denser and visual acuity decreased to 20/100 in that eye. The patient underwent orbital examination by means of an MRI scan but no distinctive signal intensity sequence was observed between both vitreous cavities.

**Discussion**

Vitreous opacification caused by amyloid was first reported by Kantarjian and DeJong [4] in 1953 in cases of neuropathic familial amyloidosis. Subsequently it has been proved that they appear only in several TTR-dependent forms of amyloidosis. A variant TTR with one aminoacid substitution of methionine for valine at position 30 is associated with type I FAP. This is an autosomally dominant inherited disorder with incomplete penetrance and characterized by polyneuropathy and autonomic dysfunction as the prominent indications. Vitreous deposits are present in about 10% of all cases of FAP I [2]. They have also been found to occur in FAP type II (TTR-Ser-84)[5], the Jewish FAP type (TTR-Ile-33) [6] and, recently, in a new TTR mutation (TTR-Asp-84) [7]. Our patient carries the TTR-Met-30 mutation characteristic of FAP type I.

Some papers describe cases of FAP where there is vitreous amiloidosis but no family history of it [8–12]. Others report cases of a singular occurrence of the ocular symptoms and vitreous deposits without neuropathy, but generally where other cases of FAP exist in their relatives [3]. In our patient both the absence of a family history of the disorder and the single ocular manifestation coincided. Reports on similar cases are rare in the ophthalmic literature. Ferry and Lieberman [11] in 1976 reported on the case of a 59-year-old woman with amyloidosis of the vitreous body without familial or systemic involvement over a seven-year period. Subsequently similar cases have been reported but, generally, in patients coming from the northern part of Sweden, where there is a known focus of FAP type I [2, 9, 10, 13].