

NONFAMILIAL VITREOUS AMYLOIDOSIS DIAGNOSED BY PORTABLE SUTURELESS VITRECTOMY

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Purpose: To describe a case of primary nonfamilial vitreous amyloidosis and a novel technique for expediting vitreous biopsy.

Design: Interventional case report.

Case: A 43-year-old man presented with progressive deterioration of vision and was found to have bilateral vitreous opacities. A systemic medical workup including family history was noncontributory. Given a high clinical suspicion of vitreous amyloidosis, the decision was made to obtain a vitreous biopsy for ultrastructural study. An office-based pars plana sutureless vitrectomy was performed. Pathologic study of the vitreous specimen confirmed the diagnosis of amyloidosis.

Conclusion: Vitreous amyloid deposition may occur with neither systemic involvement nor family history. Sutureless pars plana vitrectomy may facilitate diagnosis while saving time and expense for both the physician and the patient.

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Vitreous amyloidosis is an unusual cause of vitreous opacity. Whereas primary familial ocular amyloidosis is well documented, primary nonfamilial vitreous amyloidosis remains exceedingly rare, with only 12 cases identified in the ophthalmologic literature. Diagnosis depends on a high index of clinical suspicion and a specimen for pathologic study. To our knowledge, here we describe the first case of primary nonfamilial vitreous amyloidosis diagnosed by portable, sutureless, pars plana vitrectomy.

Case Report

A 43-year-old man of Mexican descent presented with a 6-month history of progressive bilateral decrease in visual acuity. The patient reported periocular pain and denied any neurologic deficits. Several ophthalmologists had evaluated the patient before presentation to our institution.

The patient had an ocular history significant for a pterygium

Dr. Josephberg has a financial interest in the portable sutureless vitrector.

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excision in Mexico 10 years earlier. He otherwise denied any medical or surgical history. He used neither systemic nor topical medication. He reported no family history of any disease phenomenon.

At examination, visual acuity was counting fingers at 2 ft in the right eye and 20/100 in the left eye. Pupils were round and reactive without a relative afferent defect. Applanation tonometry measurements were 16 mmHg bilaterally. Results of anterior segment examination were normal, and extraocular muscle movements were full in both eyes. Dilated fundus examination revealed consolidated “glass wool” vitreous opacities in both eyes (Fig. 1) with a normal appearing retina and optic nerve. B-scan ultrasound examination confirmed the clinical finding of vitreous densities. Subsequent fluorescein angiography was significant for bilateral vascular tortuosity without leakage and an obstructed view of the posterior pole.

A presumptive diagnosis of vitreous amyloidosis was made based on the clinical findings, and the patient was referred to the medical department for a systemic workup for amyloidosis. The patient was also petitioned to explore his family’s medical history for any polyneuropathy or unexplained illness. At follow-up, results of the systemic workup were negative, and the family history remained noncontributory.

The decision was made to acquire a vitreous specimen for pathologic analysis. With the aid of a slit lamp, a minicore biopsy was performed using a battery-powered vitrector. Specifically, a 23-gauge self-sealing transconjunctival portable vitrector with indwelling cutter and aspirator was inserted at the pars plana of the left eye and directed toward the vitreous opacities. Approximately 0.7 mL of vitreous was obtained and received on ice at the New York Eye and Ear Infirmary for review by an ophthalmic pathologist.

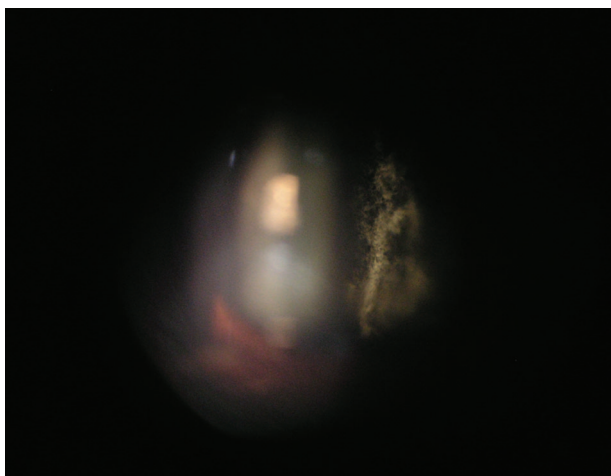


Fig. 1. Vitreous appearance by slit-lamp examination.

Congo red staining of the sample displayed coarse fibrillar aggregates (Fig. 2), which under polarized light displayed dichroic apple green to orange birefringence, consistent with amyloid. In the absence of any family history and no systemic manifestation of amyloidosis, the diagnosis of nonfamilial primary vitreous amyloidosis was made.

Discussion

Amyloidosis represents an array of disease defined by the extracellular deposition of the insoluble amyloid fibril. Deposition may be local or systemic and uniformly consists of fibrils with a β -pleated sheet secondary structure. The amyloidoses are classified by the origin of the fibril's precursor protein. The hereditary amyloidoses describe those types of amyloid that are inherited in an autosomal dominant pattern.

Among the hereditary amyloidoses, the accumulation of variants of transthyretin occurs most frequently, commonly manifesting with a progressive polyneuropathy.¹ The first description of familial amyloidotic polyneuropathy was from Portugal in 1952.² Currently, familial amyloidotic polyneurop-

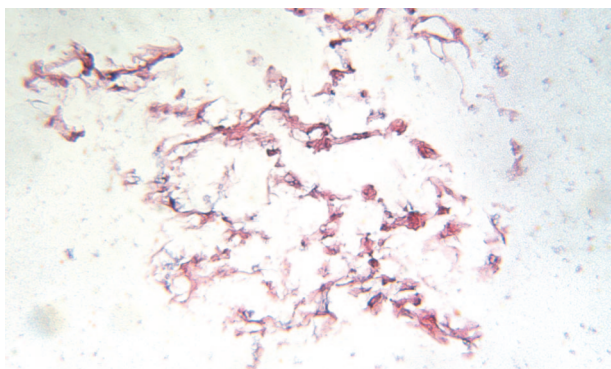


Fig. 2. Congo red staining of the sample (magnification, $\times 40$).

athy is characterized by the systemic deposition of mutated transthyretin. Most cases of mutated transthyretin in familial amyloidotic polyneuropathy are caused by the amino acid substitution of methionine for valine at position 30.

Amyloid deposition in the vitreous is typically a manifestation of a neuropathic familial amyloidosis and is generally associated with a responsible mutation of transthyretin. Far more infrequently, vitreous amyloid deposition has been described with neither systemic amyloid deposition nor family history of disease. In the review by Sandgren² of vitreous amyloidosis, only 10 cases of nonfamilial vitreous amyloid without neuropathy were identified. Since then, only two similar cases have been reported.³ The average age in these 12 cases was 61.6 years.

Vitreous amyloidosis, regardless of etiology, typically presents with progressively decreasing bilateral or unilateral vision. At examination, the vitreous opacities typically have a "ground glass" or "cobweb" appearance. Diagnosis is dependent on a high degree of clinical suspicion and tissue biopsy. Specimens are stained with Congo red dye and viewed with polarized light, revealing apple green birefringence.

To date, vitreous biopsy necessitated pars plana vitrectomy. However, as illustrated herein, a sufficient amount of vitreous may be biopsied using an office-based sutureless vitrector. Portable vitrectomy has advantages for both the clinician and the patient. Without the need to schedule operating department time, cost is minimized for the patient and a diagnosis may be expedited. For example, in our case a pathologic diagnosis was made within 24 hours of the decision to obtain a biopsy specimen. The 23-gauge apparatus described herein makes the same size scleral opening as the 25-gauge trocar systems commonly available on the market but is not limited to use in the hospital's operating room or ambulatory surgery setting. Sutureless pars plana vitrectomy can be useful for diagnostic vitrectomy, although standard therapeutic vitrectomy may be required after diagnosis is established.

Acknowledgment

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References

1. Kasper DL, Braunwald E, Fauci AS, et al. *Harrison's Principles of Internal Medicine*. 16th ed. New York: McGraw-Hill; 2004.
2. Sandgren O. Ocular amyloidosis, with special reference to the hereditary forms with vitreous involvement. *Surv Ophthalmol* 1995;40:173-196.
3. Knapp CM, Sarodia U, Brown L, Bibby K. Primary nonfamilial ocular amyloidosis. *Eye* 2003;17:252-254.