Clinical Findings in a Choroideremia Patient Who Underwent Vitrectomy for Retinal Detachment Associated with Macular Hole

Choroideremia is an X-linked, hereditary disorder characterized by progressive degeneration of the choroid, retinal pigment epithelium (RPE), and retina. The responsible gene, CHM, encodes Rab escorting protein 1 (REP-1), which relates to intracellular vesicular transport.1 Retinal detachment associated with macular hole develops predominantly in highly myopic eyes in the presence of tangential macular traction caused by vitreous degeneration.2 Here, we describe the clinical findings in a choroideremia patient who suffered from retinal detachment associated with macular hole. The unique intraoperative findings may improve our understanding of the pathological conditions of choroideremia.

Case Report

A 40-year-old Japanese man presented in October 2008. He already had impaired visual acuity due to choroideremia, which had been clinically diagnosed as chorioretinal atrophy (Fig. 1A, B). He presented because he became aware of further visual loss in his left eye (best-corrected visual acuity, BCVA, 0.04 OS, 0.2 OD). Examination showed that the cause was retinal detachment associated with macular hole.
Figure 1A–G. Preoperative findings. A, B Preoperative fundus photographs. Retinal pigment epithelium atrophy was observed in both eyes except in the area around the optic disc. The macular hole (arrows in B) and retinal detachment are shown. C Optical coherence tomography (OCT). Macular hole, retinal detachment, and a thin retina are observed. A single-flash full-field electroretinogram (D) and Goldmann visual field analyses (E, F) showed changes typical of choroideremia. G Nucleotide sequences around the mutation in the CHM gene. The vertical arrow shows the nucleotide position where C was replaced by T.

(Fig. 1B). This was confirmed by optical coherence tomography (OCT), which further revealed a very thin retina (Fig. 1C). He had −10 diopters of high myopia (axial length: left, 26.7 mm; right, 27.0 mm). In addition to a flat-type electroretinogram (Fig. 1D) and typical visual field defects (Fig. 1E, F), a substitution of C for T at nucleotide position 838 (838C→T) in the CHM gene (hemizygous R270X) confirmed the diagnosis of choroideremia (Fig. 1G).

Surgery, pars plana vitrectomy with a 23-gauge microincision, was performed in December 2008. During the vitrectomy we found advanced vitreous liquefaction and a lower than normal amount of vitreous gel in the vitreous cavity. Surprisingly, there was no residual vitreous on the posterior retina and no vitreous schisis, confirmed by visualizing the posterior hyaloid with an intraocular injection of triamcinolone acetonide, suggesting that severe tangential tractional force on the retina was absent.

The only tissue we could peel from the retinal surface, guided by indocyanine green dye, was a cord-like transparent structure that might have been part of the internal limiting membrane (ILM), but it was far thinner and tore more easily than the ILM usually seen in highly myopic eyes. After gas