Macular Changes in Type I Gaucher’s Disease

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Abstract. The authors illustrate the spectrum of Gaucher’s disease involving the eye in the case of a 51-year-old man suffering from Type I Gaucher’s disease who presented with unusual macular changes. This is the first report of chronic adult non-neuronopathic disease (Type I) with a plaque-like mass at the fovea. Our hypothesis is that the lesion at the fovea is probably an aggregation of Gaucher’s cells. [Ophthalmic Surg Lasers 2000;31:331-333]

INTRODUCTION

Gaucher’s disease is a rare familial disorder of lipid metabolism resulting in an accumulation of abnormal glucocerebrosides in the reticulo-endothelial system. It is manifested clinically by hepatosplenomegaly, skin pigmentation, and skeletal lesions. Ocular manifestations include scattered white spots in and on the retina, typical pinguecula, and supra-nuclear oculomotor apraxia in patients with Type III subacute juvenile neuronopathic Gaucher’s disease.1

CASE REPORT

A 51-year-old man suffering from Type I Gaucher’s disease (chronic adult non-neuronopathic), treated with imiglucerase injections, was referred to our center for ophthalmic assessment. The patient had no ocular complaints. His best corrected visual acuity was 6/6 in the right eye (OD) and 6/12 in the left eye (OS). Slit lamp examination of the anterior segment revealed no abnormalities and no pingueculas; intraocular pressure was normal. Fundus examination OS disclosed an elevation at the fovea (Figure 1).

Amsler grid test was normal. Humphrey visual field test revealed a relative central scotoma OS. B-scan ultrasonography (Figure 2) detected an echo-dense plaque-like lesion with shadowing effect in the center of the macula. Fluorescein angiography OS showed a hypofluorescent lesion at the fovea with annular hyperfluorescent staining. This lesion did not change in size over time (Figure 3).

Heidelberg retinal tomography shows an area of elevated macula of 2.2 mm2, elevated volume of 0.2 mm3, maximal height of 0.1 mm above the retinal surface, and the maximal diameter of the elevated macula of 1.8 mm (Figure 4). Indocyanine green demonstrates retinal pigment epithelium elevation with juxtafoveal hot spot and hypofluorescent at the upper margin (Figure 5).
COMMENT

Gaucher's disease is a rare autosomal recessive disorder of lipid metabolism resulting in an accumulation of abnormal glucocerebrosides in reticuloendothelial cells. It is manifested clinically by hepatosplenomegaly, skin pigmentation, and skeletal lesions. The disease is characterized by a lack of glucocerebrosidase activity, which normally hydrolyzes glucocerebroside to glucose and ceramide, resulting in accumulation of glucocerebroside.² The use of an analogue of the human enzyme produced by recombinant DNA technology, catalyzes the hydrolysis of the glycolipid glucocerebroside to glucosyl ceramide.

The characteristic ophthalmological manifesta-
tions of subacute juvenile neuropathic Gaucher's disease (Type III) are scattered white spots in and on the retina, retinal hemorrhages and edema, a solitary posterior pole lesion with a pigmented temporal margin, granular opacities in a ring-shaped distribution around the fovea, and macular cherry-red spots. Typical pinguecula and the supra-nuclear oculomotor apraxia are also found in Type III. These white spots are thought to be swollen histiocytes (Gaucher's cells) or defective healing of old minute hemorrhages.

To our knowledge, this is the first report of chronic adult non-neuropathic disease (Type I) involving the fovea. We believe that the lesion at the fovea is probably an aggregation of Gaucher's cells that have been transformed into a plaque-like mass.

REFERENCES


ERRATUM

In the May/June issue, data in figures 1D, 2E, and 3B of the article Dome-Shaped Detachment of Premacular Vitreous Cortex in Macular Hole Development, (Keisuke Mori, et al), were inadvertently omitted. They are reprinted here in their entirety.

Figure 1. (D) A perimacular circular tomogram with a diameter of 3.00 mm—note the large vitreoretinal separation in every direction.

Figure 2. (E) A vertical OCT image illustrates the intraretinal splitting and a dome-shaped vitreoretinal separation.

Figure 3. (B) A horizontal OCT section through the fovea—note a dome-shaped vitreoretinal separation with a foveolar adhesion.