

## Pars Plana Vitrectomy for Tractional Retinal Detachment in a Patient with Type 3 Gaucher's Disease: An Interventional Case Report

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**Abstract:** A 12 years old boy, diagnosed with type 3 Gaucher's Disease (GD), presented with white vitreous opacities with tractional retinal detachment in the macula. Best-corrected visual acuity was 20/40 and counting fingers at 1.5m in the right and left eye, respectively. Vitrectomy was performed to improve the retinal detachment of the left eye. The visual acuity reached to 20/60 three months after the surgery and the retina was remained attached and no opacity recurrence was noted after 1 year. Therefore, Tractional retinal detachment may be a presentation of GD. The visual prognosis may be good after vitrectomy with membrane removal.

**Keywords:** Gaucher disease; Epiretinal membrane; Tractional retinal detachment

### 1. INTRODUCTION

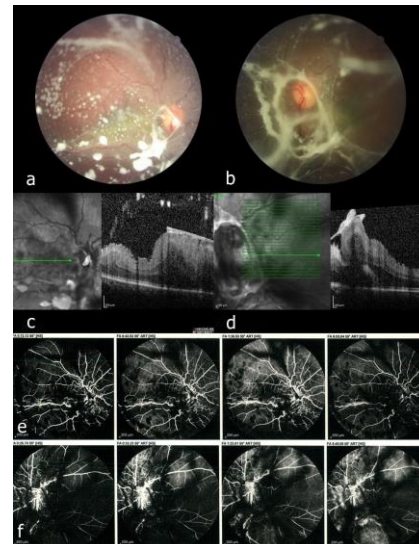
Gaucher disease (GD) is a rare autosomal recessive storage disease caused by malfunctioning of the catabolic enzyme  $\beta$ -glucocerebrosidase.[1,2] Ocular manifestations such as corneal clouding, vitreous organization, epiretinal membrane, peripheral retinal degeneration, and total tractional retinal detachment have been reported in patients with GD.[2, 3, 4] Here, we present a case of GD type 3 with macular involving tractional retinal detachment.

### 2. REPORT A CASE

The patient was a 12 years old boy, diagnosed with type 3 GD since 6 years ago. He was referred to our clinic complaining of blurred vision, mostly in his left eye, that had been significantly deteriorating during the last 6 months.

At the examination, best-corrected visual acuity was 20/40 (Spherical equivalent: +1.75 diopters sphere) and counting fingers at 1.5m (Spherical equivalent: +2.5 diopters sphere) in the right and left eye, respectively. Slit-lamp examination of the anterior segment and the intraocular pressure were unremarkable. Funduscopy revealed numerous dense whitish opacities of varying sizes overlying the optic nerve head and vessels in both eyes, more prominent in the left eye. These white opacities were also abundant on the surface of the retina. In the left eye, there were thick epi-retinal membranes around the optic disc

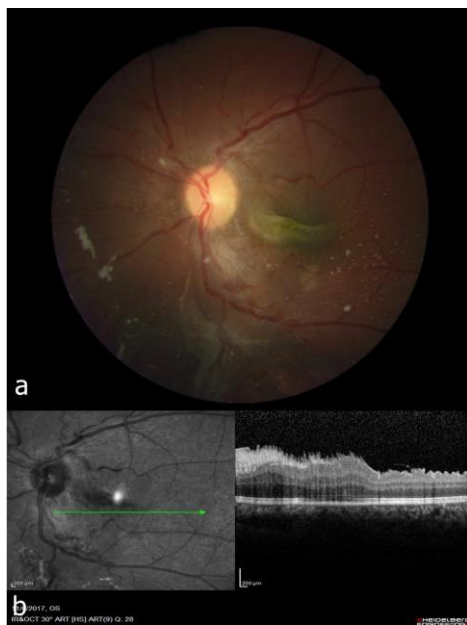
that caused tractional retinal detachment in nasal part of the macula. (Figure1) Arterial and venous tortuosity was seen in both eyes with no obvious leakage in fluorescein angiography.



**Figure1.** (a,b) fundus photos of both eyes reveal numerous dense whitish opacities of varying sizes overlying the optic nerve head and vessels, more prominent in the left eye. Arterial and venous tortuosity is seen in both eyes. (c,d) Optical coherence tomography (OCT) reveals hyper-reflective opacities adjacent to the retina surface in the vitreous cavity. Epiretinal membrane with macular pucker is evident in the right eye. An epi-retinal membrane causing a tractional retinal detachment in papillomacular area is also noted in the left eye. (e,f) Multiple hypofluorescent areas due to blockage are evident in fluorescein angiography of both eyes with no vascular leakage.

Twenty-three-gauge vitrectomy was performed to manage the retinal detachment in the left eye. After pars plana core vitrectomy, due to the firm adhesion between vitreous and retina, unsuccessful attempts were repeated to separate posterior vitreous face by aspiration with a vitreous cutter. Vitreous forceps was therefore used to pull the opaque posterior vitreous cortex away from the retina to create posterior vitreous detachment (PVD). The thick membrane was peeled from the retina along with posterior vitreous cortex and finally, triamcinolone was injected to reveal any remnant of the membrane.

One week after the operation, despite a persistent subretinal fluid and a few preretinal white opacities, the visual acuity was improved to 20/160. Three months after surgery, the visual acuity reached to 20/60. (Figure2) During 12 months' follow-up, the retina still remained attached and no opacity recurrence was noted.



**Figure2.** Three months after surgery: (a) the vitreous opacities are reduced significantly. (b) The thickness of retina is reduced while there is no obvious traction on it based on Optical coherence tomography (OCT) findings.

### 3. DISCUSSION

We reported a case of GD with bilateral vitreous opacities accompanied by an epiretinal membrane with macular pucker in the right eye and localized tractional retinal detachment (TRD) that involved nasal part of the macula in the fellow eye. Vitrectomy was done for the left eye due to severe visual loss. Previous studies suggested that vitreous organization occurs only in splenectomized patients.[1] This may be due to increased levels of circulating glucosylceramide

which lead to deposition in unusual organs such as the eye.[1] Our patient had not been undergone splenectomy. Recently, vitreous depositions have been reported in patients with GD that have never had a splenectomy. [2-4] Therefore, the exact pathophysiologic mechanism of glucosylceramide deposition in the vitreous cavity is unclear and it may be due to interactions between multiple factors such as genetic. The incidence of vitreous opacities was found to be 3% in a series of 80 patients with GD.5 In our case, opacities appeared despite receiving enzyme replacement therapy for about 6 years. As Coussa et al. explained, this may be related to the large molecular size of recombinant enzyme, which prevents it from crossing the blood-retina barrier.[6]

Sheck et al. illustrated that these opacities are pre-retinal, located at the vitreoretinal interface, and associated with localized PVD rather than the vitreous body.[7] We showed in our case that although the opacities are concentrated near the retina, they also present in the vitreous body.

During surgery, liquefaction of vitreous with incomplete PVD was obvious which is not common in young patients.[8] However, strong adhesions between the vitreous cortex and some parts of the retina around the optic disc were prominent. In one report of the use of vitreous surgery to treat epi-macular membrane in Gaucher disease, liquefaction of the vitreous body and PVD were shown to be advanced due to vitreous opacities, causing the vitreous body to be contracted anteriorly.[3] The firm adhesions between the cortical vitreous and the retina due to the patient's age may cause tractional retinal detachment following anterior contraction of the vitreous body.[8] Watanabe et al. recently reported a case of total tractional retinal detachment in GD and suggested a similar mechanism for retinal detachment.[5] As retinal detachment in our case was partial, we could clearly show the traction forces by OCT in both eyes.

During 3 months' follow-up, the retina was attached and the vision was significantly improved. There was no recurrence of the white opacities in vitreous body. Rapizi et al. have reported recurrence of opacities in periphery of the retina during 12 months after vitrectomy, despite improvement of the vision. In contrast to our study, they only had removed the core vitreous and left the periphery.[3] However, longer follow up period might be needed to evaluate the recurrence.

To the best of our knowledge, this is the second published report on the occurrence of retinal detachment in GD and the first case of localized tractional retinal detachment that involved the macula.[5] Further studies and future reports may explain the exact mechanism of vitreous involvements and retinal detachment in Gaucher disease.

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