

# Retinal detachment in a boy with Gaucher disease

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## Dear Editor,

I am Dr. Fang Fang, from the Department of Ophthalmology of the Second Xiangya Hospital of Central South University in Changsha, Hunan Province, China. I write to present a case report of retinal detachment with Gaucher disease (GD).

GD is a chronic, progressive, autosomal recessive lipid-storage disease caused by mutations in the lysosomal enzyme  $\beta$ -glucosidase (glucocerebrosidase), normally present in macrophage lysosomes. The impaired activity of enzyme glucosylceramidase leads to accumulation of glucosylceramide in scavenger macrophages and affected organs of the reticuloendothelial system including liver, spleen, and bone marrow<sup>[1]</sup> subsequently. The pathognomonic of the disease is enlarged macrophages with a foamy cytoplasm, which is called Gaucher cells. Three clinical forms (types 1, 2 and 3) of the disease have been recognized traditionally based on the severity of manifestations. Here we presented one GD case successfully diagnosed in the ophthalmology clinic and described the progression of ocular abnormalities in this disease.

## Report of A Case

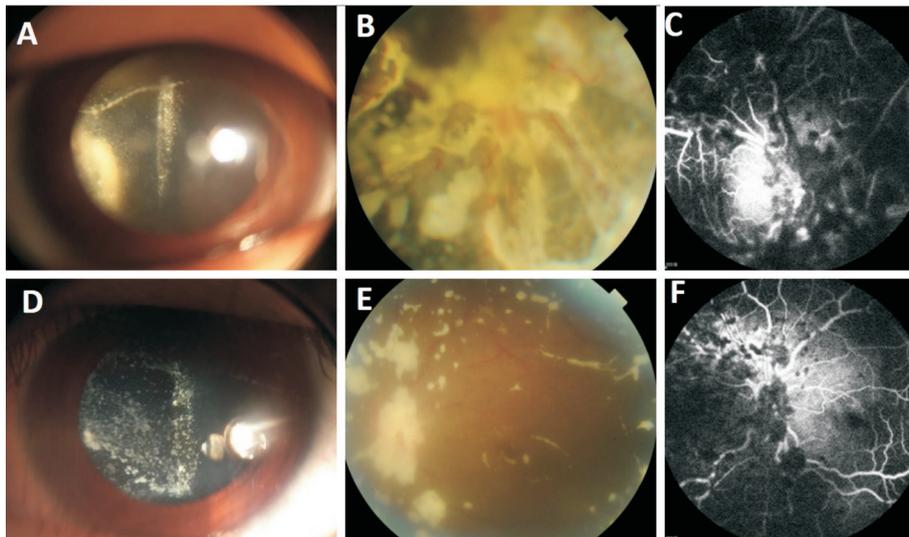
A 14-year-old boy visited our hospital complaining of blurred vision in his both eyes for two weeks. He did not exhibit obvious mental retardation. The patient had no other positive history except splenectomy due to splenomegaly at the age of six. His best-corrected visual acuity (BCVA) two years ago was 20/30 right eye and 20/20 left eye and fundus examination indicated vitreous opacities in both eyes. However, the BCVA decreased to 20/400 in both eyes at this time. No abnormalities can be captured in the anterior segment under

slit-lamp examination. There was a conspicuous absence of inflammatory cells in the vitreous bodies bilaterally. The posterior segment examination showed moderate to dense whitish vitreous dots in overlying optic discs, vessels, and vitreous bodies of both eyes and even involvement to the posterior capsule of the lens, which was worse in the right eye. The dots varied in size, some with fluffy appearance. B-scan demonstrated vitreous opacity in both eyes and retinal detachment in right eye. Fluorescein angiography (FFA) of both eyes in transit phase revealed normal filling time, blockage of fluorescence caused by vitreous opacities and marked vascular tortuosity (Figure 1). Based on all these manifestations, type 3 GD was highly suspected, which was proved by Gaucher cells found in the bone marrow specimen. The diagnose was further confirmed by the finding of homozygosity for the common p.Leu483Pro mutation in the *GBA* gene thus suggesting type 3 or neuronopathic disease (Figure 2). The patient refused to receive a surgery against the retinal detachment and any other treatment. Ten months later, the whitish dots increased significantly, the retinal detachment became much worse in both eyes and neovascular vessels started to appear in the right eye (Figure 3).

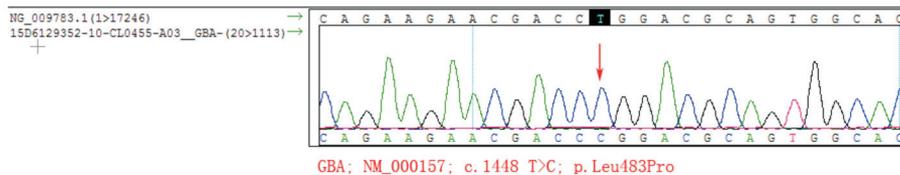
## DISCUSSION

Gaucher's disease is an autosomal recessive disorder due to deficiency of glucocerebrosidase. This reticuloendothelial storage disorder has been clinically divided into 3 subtypes based upon the absence (type 1) or presence (types 2 and 3) of neurological impairment of central nervous system<sup>[2]</sup>. The prevalence of GD in the general population has been estimated to be about 1:60 000. Classic descriptions of GD often include multiple ocular associations, such as conjunctival pterygia, strabismus, and supranuclear gaze abnormalities<sup>[3]</sup>. Vitreous opacities, one of the common ocular manifestations, were firstly reported by Cogan *et al*<sup>[4]</sup>. According to the previous reports, vitreous opacities were more inclined to occur in the GD patients undergoing splenectomy, which may due to increased circulation of glucosylceramide<sup>[5]</sup>. Watanabe *et al*<sup>[6]</sup> recently found severe vitreous opacities in both eyes of the patient without splenectomy.

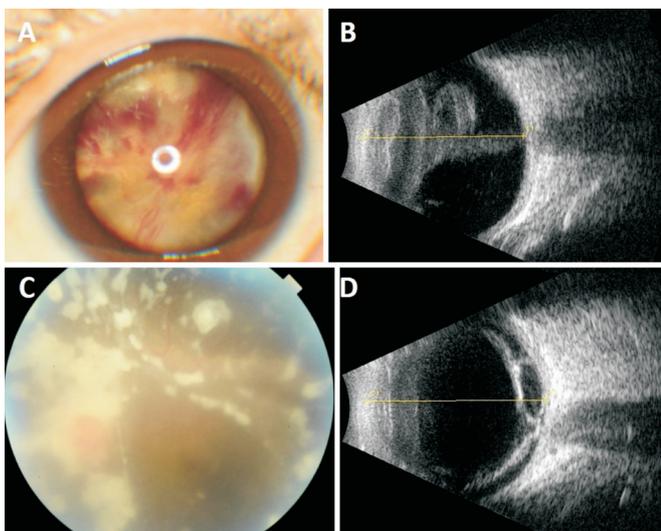
Shrier *et al*<sup>[7]</sup> and Fujiwaki *et al*<sup>[8]</sup> reported that the posterior vitreous opacities had a large amount of glucosylceramide in the vitreous specimen confirmed by pathology. However, the pathophysiologic mechanism of glucosylceramide deposition in vitreous cavity remains elusive. As a matter of fact, the



**Figure 1** The anterior segment and fundus examinations during the patient's first visit in eye clinic A-C: Right eye; D-F: Left eye. Slit-lamp examination revealed no abnormalities of cornea and anterior chamber but numerous whitish dots were attached to the posterior lens capsule and in the vitreous cavity in both eyes (A, D). Fundus photographs showed prominent whitish pre-retinal and vitreous opacities and retinal detachment in the right eye (B) and comparatively less pre-retinal and vitreous deposit overlying optic disc, vessels, vitreous in the left eye (E). Fluorescein angiography of both eyes in transit phase revealed normal filling time, blockage of fluorescence caused by vitreous opacities and marked vascular tortuosity (C, F).



**Figure 2** Location of the gene mutation responsible for the patient.



**Figure 3** The clinical findings during the follow-up after ten months The anterior segment photograph showed massive neovascular vessels and severe detached retina adhered to the posterior lens capsule in the right eye (A) which was demonstrated as a closed V-shape by the B scan (B). The fundus photograph revealed the great increase in the whitish deposit and retinal detachment in the left eye (C). Accordingly, the B scan showed obvious vitreous opacities and an extensive retinal detachment (D).

material is a by-product of breakdown of myelin, leukocytes, red blood cells, and endothelial cells. It was presumed that the vitreous opacity within the eye was induced by the migration of oligodendroglial cells through the lamina cribrosa or the infiltration of monocyte-macrophage phagocytosed system cells, which contained much glucosylceramide<sup>[9]</sup>. Besides, whether the vitreous deposits will appear again after the vitrectomy is unknown. Only one case of recurrent vitreous opacity one year after vitrectomy has been reported<sup>[10]</sup>.

To our knowledge, only one case of retinal detachment in GD was previously reported with severe vitreous opacities in a 16-year-old boy<sup>[6]</sup>. Advanced liquefaction of the vitreous body in the central part of the vitreous cavity has been noted, which rarely occurs to teenagers. The white fluffy opacities, dispersing in the vitreous cortex, strongly adhere to the retina with no tears found. Similarly, in another case with severe vitreous opacities, no retinal tear was observed during the operation and macular pucker was revealed postoperatively<sup>[7-10]</sup>. These manifestations indirectly indicate that retinal detachment in GD might be caused by mechanical disturbance of the retina by vitreoretinal traction.

In our case, although retinal detachment with vitreous opacities in the right eye is severe, no obvious retinal hole was observed. By ten months of follow-up observation without surgery, the retinal detachment deteriorated fast in both eyes, complicated with neovascular vessels in the retina of the right eye. This is the first case report that such severe vitreous opacities with retinal detachment in GD which can progressively lead to permanent loss of vision without treatment. It strongly suggests that the ocular complications of GD, such as retinal detachment, often cause blindness if without intervention. Surgery-dominated treatments are essential, which can prolong and reserve the patients' remaining visual acuity.

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**Authors' contributions:** Fang Fang and Tan-Tai Zhao wrote the manuscript and made the revisions, other authors played parts in the medical history taking and the treatment of the patient.

**Conflicts of Interest:** Zhao TT, None; Li HL, None; Guo XJ, None; Luo J, None; Liu X, None; Fang F, None.

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