Vitreous Abnormalities Of Gaucher's Disease

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Introduction

Gaucher disease (GD) is a lysosomal storage disorder caused by a recessively inherited deficiency of glucocerebrosidase activity, which caused an accumulation of glucosylceramide in various organs. Typically patients develop hepatosplenomegaly hematologic complication and osseous manifestations. Neuronopathic involvement is less common. Ocular manifestations including oculomotor apraxia, supranuclear gaze abnormalities, corneal clouding and retinal lesions have been reported. However, little is known about their natural progression. Here, we present a case with progression of vitreous abnormalities in GD.

Case report

We describe a 14-year-old girl was diagnosed at the age of 2-year-5-month old with GD Type III after presenting with hepatosplenomegaly and pancytopenia. The diagnosis was confirmed by low glucocerebrosidase activity. Genetic testing revealed homozygosity for the L444P mutation. Enzyme replacement therapy (ERT) every other week was started since diagnosis. At first her neurologic examination and fundoscopy were normal. Seizure developed at 10-year-old with antiepileptic drug control. During regular follow, hematologic problem and hepatosplenomegaly improved after ERT. At age of 14 years, color fundoscopy demonstrated the presence of much pre-retinal opacities (Figure 1). Optical coherence tomography (OCT) revealed there are no evident lesions in retina layer and many floaters in the vitreous layer (Figure 2). Visual acuity showed 20/60 in both eyes.

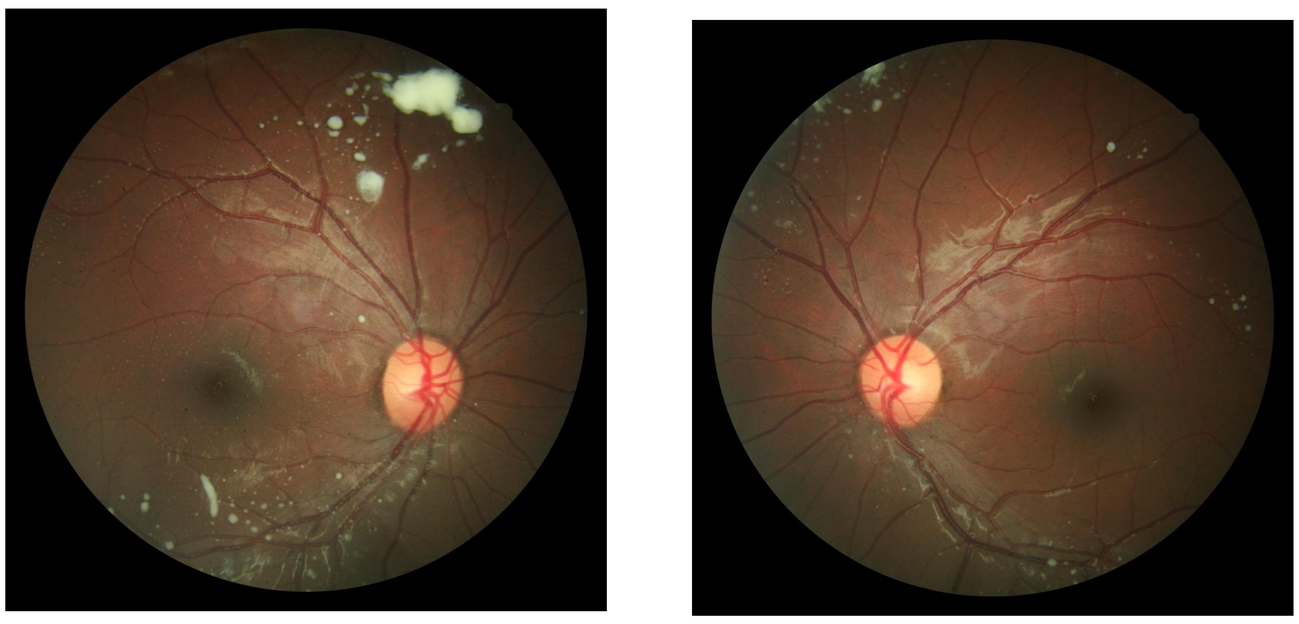

Figure 1. Color fundus photographs demonstrating the presence of much pre-retinal opacities. These opacities are enhanced on red-free imaging

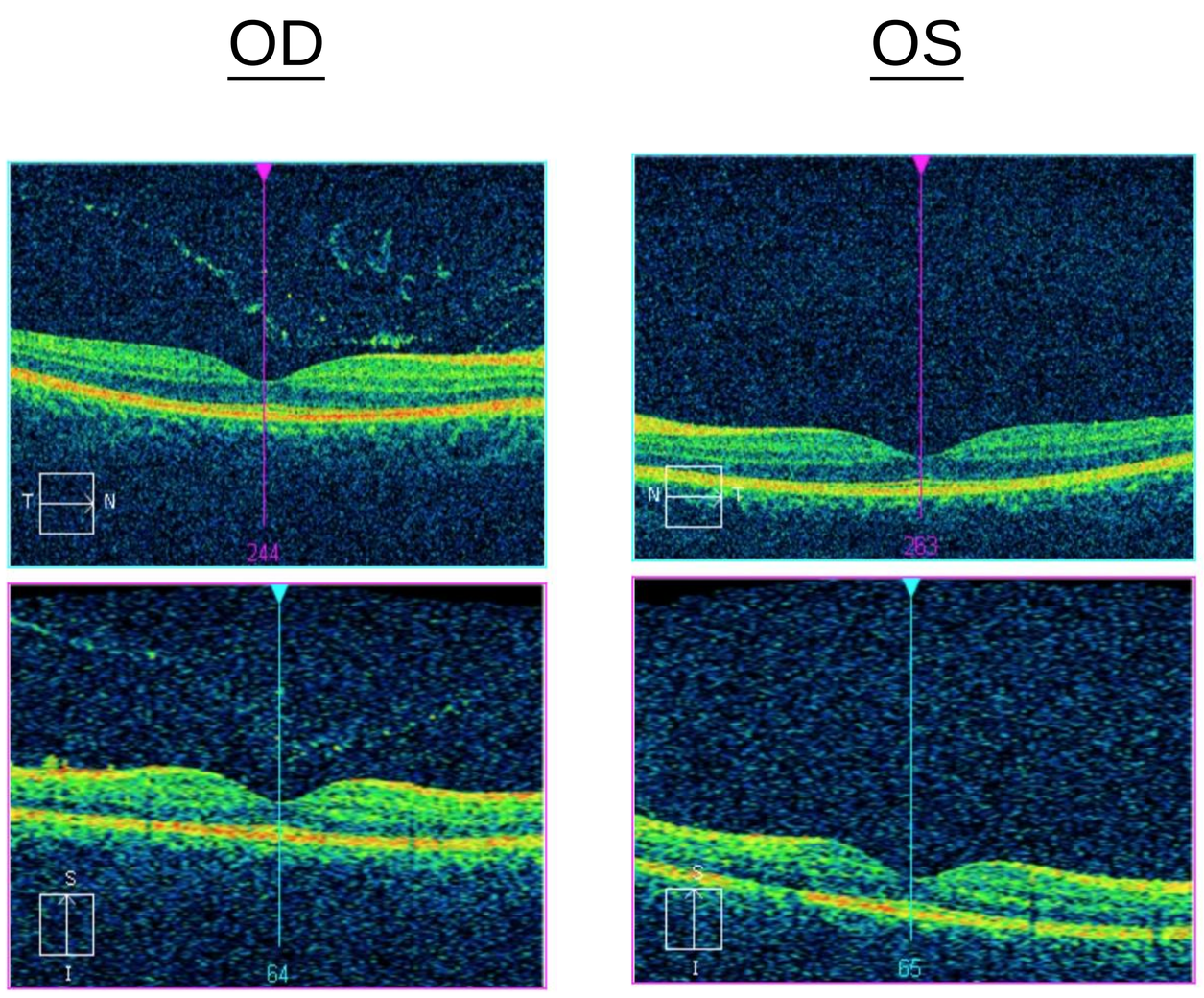

Figure 2. OU OCT revealed there are no evident lesions in retina layer and many floaters in the vitreous layer.

Discussion

Vitreous lesions have seldom reported in Gaucher patients and were thought to represent the eponymous Gaucher cells. Accumulation of higher levels of circulating glucosylceramide in unusual systemic locations has been reported [1]. In our case, the reported lesions developed after ERT for nearly twelve years. During regular follow, hematologic problem and hepatosplenomegaly improved after ERT. This finding suggested that ERT improved the systemic manifestations in patients with GD3, but seems not able to counteract the progression of ophthalmic symptoms in the long term. This possibly due to the fact that the immune-privilege enjoyed by the eye, which prevents such large proteins from crossing the blood retinal barrier [2]. The similar findings also showed in patients with neurologic symptoms. ERT for GD3, improved the systemic manifestations in patients with but was not able to counteract the progression of neurological symptoms in the long term [3].

Further studies should be enrolled to shed light on the precise nature of the ophthalmic lesions seen in Gaucher disease.

1.Seidova, S.F., et al., *Functional retinal changes in Gaucher disease.* Doc Ophthalmol, 2009. **118**(2): p. 151-4.
2.Coussa, R.G., et al., *Progression of retinal changes in Gaucher disease: a case report.* Eye (Lond), 2013. **27**(11): p. 1331-3.
3.Sechi, A., et al., *Long term effects of enzyme replacement therapy in an Italian cohort of type 3 Gaucher patients.* Mol Genet Metab, 2014. **4**(14): p. 00254-6.

Conclusion

ERT improved the systemic manifestations in patients with GD3, but seem not able to counteract the progression of ophthalmic symptoms in the long term. In this case, the reported lesions did not preventable despite ERT. This possibly due to the fact that the immune-privilege enjoyed by the eye, which prevents such large proteins from crossing the blood retinal barrier. Further studies should be enrolled.

Keywords: Gaucher disease, vitreous Abnormalities, enzyme replacement therapy