Ophthalmic Images

Posterior Segment Changes in Gaucher Disease

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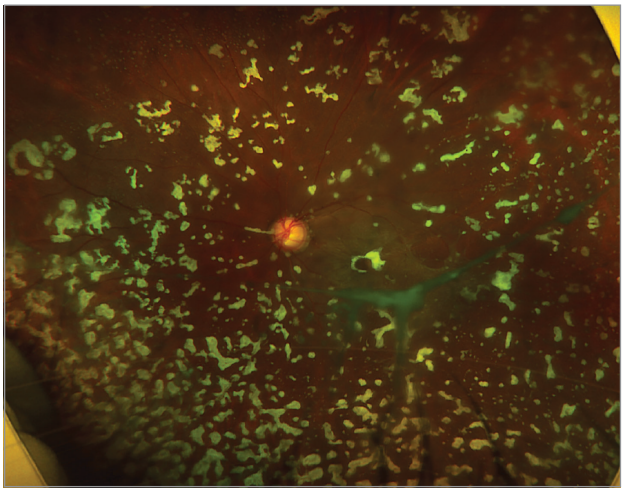
A Fundus photography



B Optical coherence tomography

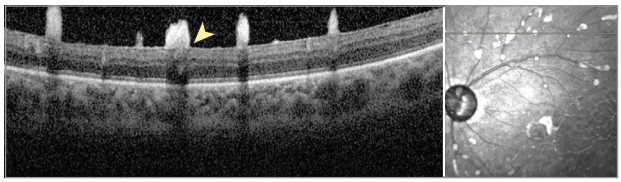


Figure. Gaucher disease-associated changes in the posterior segment of the eye. A, Fundus photography captures vitreous opacities, multiple hypopigmented spots, and significant disc cupping. B, Optical coherence tomography (OCT) demonstrates preretinal deposits (yellow arrowhead) corresponding to the observed hypopigmented spots.

A 24-year-old male diagnosed with Gaucher disease type 3 since birth presented with low vision. The patient has experienced significant complications related to the disease, including liver failure and hearing loss. Best-corrected visual acuities were no light perception OD and 20/100 OS. Open-angle glaucoma was present bilaterally with severe superior and inferior visual defects in the left eye. The right eye had a dense intumescent cataract, limiting posterior segment examination in the right eye. In the left eye, vitreous opacities and multiple discrete hypopigmented spots sparing the fovea were noted (**Figure**, A). It has been suggested that the observed vitreous opacities represent Gaucher cells, which are enlarged glucocerebroside-filled histiocytes capable of crossing the blood-ocular barrier.[1-3] The hypopigmented spots correlated with preretinal hyperreflective bodies seen on optical coherence tomography (Figure, B), likely representing glycolipid deposits.[4] Although similar findings have been previously documented,[5,6] this patient had an ultra-widefield fundus appearance with accompanying ocular coherence tomography scan showing more extensive deposits.

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ARTICLE INFORMATION

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