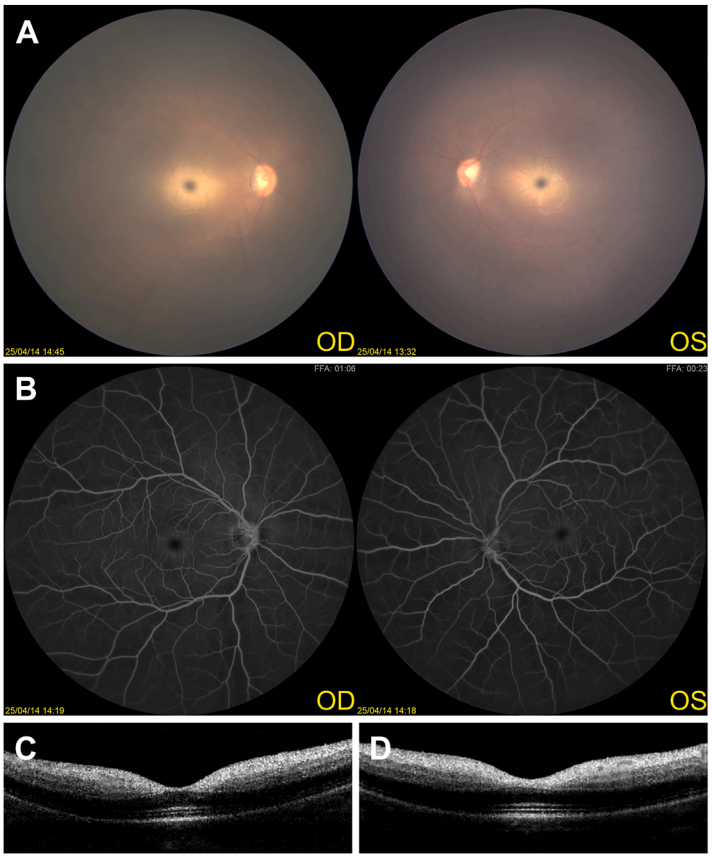
Pictures & Perspectives



Bilateral Macular Cherry-Red Spots in Infantile Sandhoff Disease

A 14-month-old boy was referred after routine ophthalmic screening detected abnormal fundus findings. Over several months, progressive developmental delay and hypotonia were noted by his parents. Widefield fundus photography demonstrated bilateral macular cherry-red spots with surrounding retinal pallor (**A**). Fluorescein angiography showed mild relative foveal hyperfluorescence without leakage or peripheral vascular abnormalities (**B**). OCT showed mild thickening of the inner retinal layers in both eyes (right eye [OD], **C**; left eye [OS], **D**). Genetic testing identified pathogenic variants in the *HEXB* gene, confirming the diagnosis of infantile Sandhoff disease. *HEXB* deficiency leads to GM2 ganglioside accumulation in retinal ganglion cells, accounting for the observed imaging features. (Magnified version of Fig **A**–**D** is available online at https://www.ophthalmologyretina.org).

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