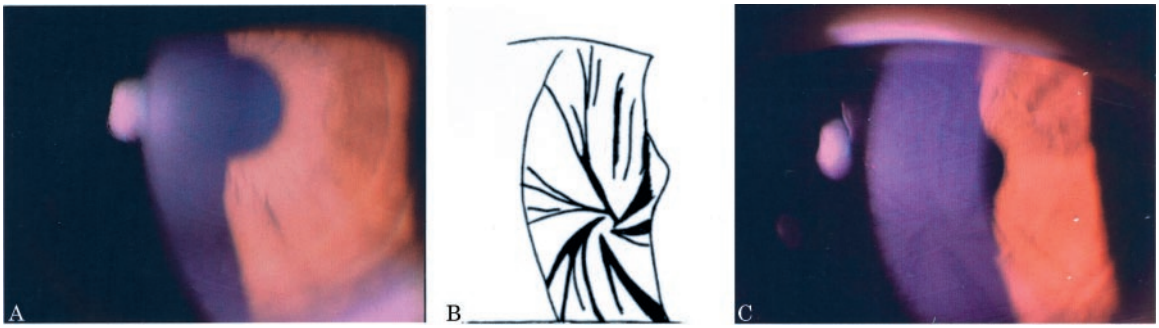

Figure. Slit-lamp examination of cornea revealed white-colored, whorl-like corneal opacity with a radial pattern. (A) Normal cornea, (B) cartoon drawing of C, and (C) the patient's current figure.

Clinical diagnosis of Fabry disease Whorl-like corneal opacity

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An 11-year-old Japanese girl preserved with eye itching and mild hypesthesia in her extremities. Her hidrosis was decreased, and her body temperature was approximately 37 °C. Family history was not contributory. Routine laboratory and physical examinations revealed no abnormal findings. Cardiac and renal function was normal. Ophthalmologic examination with a slit-lamp disclosed bilateral whorl-like corneal opacity (figure). The α-galactosidase activity in leukocytes was markedly decreased, and a point mutation of the α-*galactosidase A* gene was detected. Thus, she was diagnosed as a female carrier of Fabry disease. Whorl-like corneal opacity is a characteristic ocular manifestation of Fabry disease, even if the patient is asymptomatic or heterozygous.[1,2]

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