Neuro *Images*

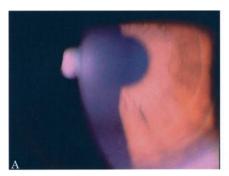






Figure. Slit-lamp examination of cornea revealed white-colored, whorl-like corneal opacity with a radial pattern. (A) Normal cornea, (B) cartoon drawing of C, and (C) the patient's current figure.

Clinical diagnosis of Fabry disease Whorl-like corneal opacity

Hideto Yoshikawa, MD and Izumi Ogawa, MD, Niigata, Japan

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. Ophthal-

Address correspondence and reprint requests to Dr. Hideto Yoshikawa, Department of Pediatrics, Niigata City General Hospital, 2-6-1 Shichikuyama, Niigata 950-8739, Japan; e-mail: hideto@hosp.niigata.niigata.jp

mologic 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

- Lou HOC, Heidensleben E, Larsen HW. The value of ocular findings in the diagnosis of angiokeratoma corporis diffusum (Fabry's disease). Acta Opthalmol 1970;48:1185–1194.
- Sher NA, Letson RD, Desnick RJ. The ocular manifestations in Fabry's disease. Arch Opthalmol 1979;97:671-676.



Clinical diagnosis of Fabry disease: Whorl-like corneal opacity

Hideto Yoshikawa and Izumi Ogawa Neurology 2003;60;1048 DOI 10.1212/01.WNL.0000048563.17902.5E

This information is current as of March 25, 2003

Updated Information & including high resolution figures, can be found at: **Services** http://n.neurology.org/content/60/6/1048.full

References This article cites 2 articles, 0 of which you can access for free at:

http://n.neurology.org/content/60/6/1048.full#ref-list-1

Subspecialty Collections This article, along with others on similar topics, appears in the

following collection(s):
All Neuro-ophthalmology

http://n.neurology.org/cgi/collection/all_neuroophthalmology

Metabolic disease (inherited)

http://n.neurology.org/cgi/collection/metabolic_disease_inherited

Permissions & Licensing Information about reproducing this article in parts (figures, tables) or in

its entirety can be found online at:

http://www.neurology.org/about/about the journal#permissions

Reprints Information about ordering reprints can be found online:

http://n.neurology.org/subscribers/advertise

Neurology ® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright . All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

