Ligneous Conjunctivitis in a Mexican Patient With a Mutation in the Plasminogen (PLG) Gene

Ligneous conjunctivitis is an autosomal recessive inherited disease caused by mutations in the plasminogen (PLG) gene. Herein, we report a girl affected by this disorder, caused by a homozygous deletion of 14 base pairs in exon 5 of the PLG gene.

Report of a Case. A 2-year-old Mexican girl, product of the fourth pregnancy of second-grade consanguineous parents with no relevant family history, was cared for at the Service of Ophthalmology of the University Hospital, Universidad Autónoma de Nuevo León, because of a refractory bilateral conjunctivitis noticed when she was 2 months of age. The physical examination revealed chronic inflammation of the eyelids and synchiae of the tarsal and bulbar conjunctivae with hard pseudomembranes in both eyes; the rest of the examination results were normal. The parents did not show any abnormality in the eyes or facial mucous epithelia. The patient required surgical removal of the pseudomembranes after failing medical treatments with topical antibiotics and corticosteroids.

The evaluation of the fibrinolytic route demonstrated very low concentration of plasmatic plasminogen (<1 mg/dL [<0.113 µmol/L]); reference range, 7-17 mg/dL [0.791-1.921 µmol/L]) and plasminogen functional activity (6%; reference range, 75%-140%), confirming the diagnosis of ligneous conjunctivitis. The patient and her parents were studied for abnormalities in the PLG gene located at 6q26. Genomic DNA were isolated from peripheral blood and DNA samples were amplified by polymerase chain reaction using different sets of primer pairs designed in the Department of Biochemistry for analysis of exons 2, 5, 7, 10, 13, 14, 15, and 17 of the PLG gene. All amplicons included exon-intron boundaries. Polymerase chain reaction products were purified and sequenced in an automated DNA sequencer (Li-Cor DNA 4200; Li-Cor Inc, Lincoln, Neb) and confirmed by restriction fragment length polymorphism analysis. This study showed a homozygous nucleotide deletion of 14 base pairs in exon 5 of the PLG gene in the patient, which creates a stop codon at position 145 of the protein. The deletion eliminates a restriction site for PsI present in the wild-type allele. After digestion with PsI (New England Biolabs, Inc, Ipswich, Mass), amplicons were electroforesed in 2.5% agarose gel and visualized by ethidium bromide staining. Both parents were heterozygous for the mutation (Figure).

Comment. Ligneous conjunctivitis is a rare disorder characterized by re-

![Figure](https://example.com/figure.png)

©2006 American Medical Association. All rights reserved.

Downloaded From: https://archopht.jamanetwork.com/ by a Non-Human Traffic (NHT) User on 03/07/2019
current lesions of hard consistency in the mucous membranes, which have a negative effect on homeostatic fibrinolysis because of mutations in the PLG gene. This disorder is usually detected in young children with recurrent conjunctivitis due to pseudomembranes that evolve to hard nodular masses of woodlike consistency.1 Ligneous conjunctivitis has an autosomal recessive pattern of transmission and several mutations in the PLG locus have been demonstrated in patients with this disorder.1 A mutation screening performed on the patient and her parents demonstrated a nonreported, homozygous, 14-base pair deletion in exon 5 of the PLG gene, which generates an early stop in the 5′ region of the gene. One recent report suggests a possible “hot spot” region around the Cys133-Cys157 in the PLG gene.2 We hypothesize that this homozygous mutation results in the synthesis of a truncated plasminogen molecule in the liver that is quickly degraded,3 a finding that correlates with the early onset and the severity of the eye abnormalities. Despite this physiologic defect, it is noticeable that the patient does not show involvement in other organs besides the eyes.

Juan M. Leyva-Castillo, QFB Humberto Cazazos-Adame, MD Augusto Rojas-Martinez, DSc, MD Roberto Lopez-Garza, MD Martin Cesar Fernandez Espinoza, MD Rocio Ortiz-Lopez, DSc

Correspondence: Dr Ortiz-Lopez, Department of Biochemistry, School of Medicine, Universidad Autonoma de Nuevo Leon, Ave Francisco I. Madero y Dr Aguirre Pequeño, Monterrey, Nuevo Leon, CP 64460, Mexico (rortiz@fm.uanl.mx).

Financial Disclosure: None reported.

Funding/Support: This work is supported by the Departments of Biochemistry and Ophthalmology, School of Medicine and University Hospital, Universidad Autonoma de Nuevo Leon.


Melioidosis With Endophthalmitis

Melioidosis is an infectious disease caused by Burkholderia pseudomallei, a Gram-negative bacillus. It is endemic to southeast Asia and northern Australia as well as regions between 20° latitude north and south of the equator. The clinical manifestation varies from a latent infection with an incubation period of up to 29 years to fulminant sepsis with a high mortality rate.1 We report a case of endogenous endophthalmitis caused by B pseudomallei with a fulminant course.

Report of a Case. A 70-year-old Chinese male veteran living in southern Taiwan had a medical history that included controlled diabetes mellitus for more than 10 years and coronary artery disease after bypass graft surgery 2 years previously. Ocular history included herpetic keratouveitis in the right eye 4 years previously with residual corneal opacity, bilateral senile cataract, and bilateral nonproliferative diabetic retinopathy. Ocular trauma and operation history were not reported. At initial examination on August 5, 2005, the patient had pain in the right eye and headache that had persisted for 1 day. Visual acuity of the right eye was decreased to light perception. Marked chemosis, corneal edema, hyphema, and elevated intraocular pressure were found. He was diagnosed with and treated for neovascular glaucoma. On day 11, hypopyon and a localized scleral suppurition, which disseminated over the following days (Figure 1), were noted. The vitreous echoes were heterogeneous with a fluffy retinal surface. Vitreous fluid was aspirated for smear and culture. Gram-staining smear disclosed numerous Gram-negative rods (Figure 2). The diagnosis of endogenous endophthalmitis was made. After immediate intravitreal injection of 1 mg of vancomycin hydrochloride, 0.4 mg of amikacin sulfate, and 0.4 mg of dexamethasone sodium phosphate, topical eyedrops consisting of vancomycin hydrochloride (50 mg/mL) and amikacin sulfate (25 mg/mL) were administered in combination with 4 g/d of intravenous ceftriaxone sodium. On day 16, both vitreous and blood cultures yielded B pseudomallei. Systemic cefazidine, cotrimoxazole, and granulocyte colony-stimulating factor were administered.1 However, the illness progressed to septic shock and multiple organ failure, and the patient died on day 18. Systemic survey

Figure 1. The right eye on day 18 showing disseminated multifocal scleral suppuritation.