

Case Report

Corneal Opacities in a Case of Leber Congenital Amaurosis due to Excessive Eye Rubbing

Christoph Faschinger*, Susanne Lindner, Dieter Franz Rabensteiner

Department of Ophthalmology, Medical University Graz, Austria

***Corresponding author:** Christoph Faschinger, Department of Ophthalmology, Medical University Graz, A-8036 Graz, Austria. Tel: +43-31638582899; Email: Christoph.faschinger@medunigraz.at

Citation: Faschinger C, Lindner S, Rabensteiner DF (2019) Corneal Opacities in a Case of Leber Congenital Amaurosis due to Excessive Eye Rubbing. Case Report. Ann Case Rep: ACRT-213. DOI: 10.29011/2574-7754/100213

Received date: 30 December 2018; **Accepted Date:** 24 January 2019; **Published Date:** 01 February 2019

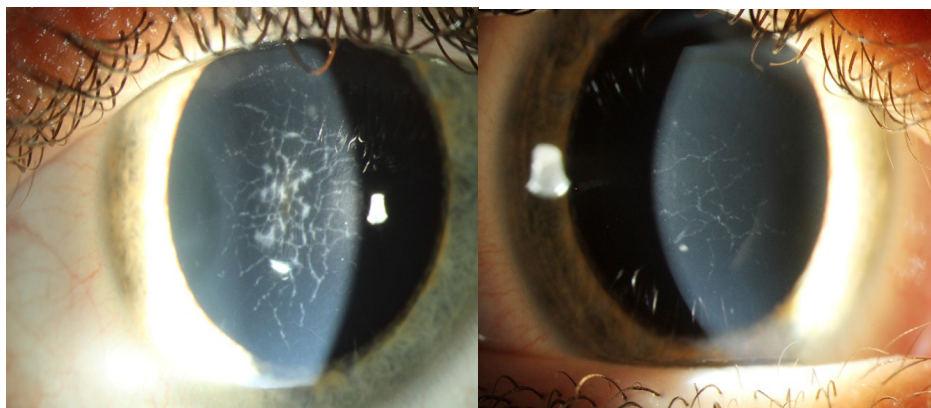
Case Report

J.G. was born via Caesarean section in 2002 as the first child of a 30-year old mother after a completely uneventful pregnancy. After a few weeks the parents recognized that the boy did not fixate any objects and had developed a horizontal nystagmus with strange oscillations. An eye examination of both eyes revealed no obvious inflammations, a clear cornea, a clear lens, no vitreous opacities, but a pale optic nerve head. Both pupils reacted sluggishly to light. A proposed cerebral Magnetic Resonance Imaging (MRT) examination was refused by the mother. The diagnosis Leber Congenital Amaurosis (LCA) was suspected. An early professional visual stimulation was initiated. During the following years the visual acuity never improved to more than positive light perception in both eyes. The discs remained pale, the macula showed a soft granular pattern and the periphery looked like salt and pepper, the retinal vessels were thinner than normal.

The boy went to school, finished elementary school and junior high school and was very skilful in working with the computer and Braille software. Against blur and photophobia, he wears polarized glasses with filters blocking light beyond 550 nm. For navigation he uses a white cane. Balance and coordination are functioning well. To confirm the diagnosis a recent genetic analysis of the boy and the mother was performed (the father of the boy refused to be tested). Both, mother and son carried the CEP290 mutation c.4990G>T, a stopcodon on chromosome 12 (12q21.32) and was heterozygote in each case. In contrast to the

son, the mother showed no CEP290 mutation c.4723A>T. An autosomal recessive LCA type 10 was confirmed [1]. The genetic analyses were performed by the Department of Medical Genetics, Molecular and Clinical Pharmacology (Human Genetics Division, Head: J Zschocke, MD, PhD) Innsbruck in 2018.

Because the mother discovered white changes in the cornea of both eyes, they went to see an ophthalmologist additionally to the annual check-ups. In both eyes (right >> left) the corneas showed superficial white irregular shaped netlike crossing lines, some of them being a little bit thicker (Figures 1,2). Upon questioning, the mother confirmed that the son was still rubbing his eyes (oculodigital phenomenon). The start of this eye rubbing is documented in the boy's medical charts in his 10th week of life. He continued to do so for many years until today, sometimes still with his knuckles. According to the denser amount of the whitish line-like changes in his right eye, he probably does it more intensively with his right hand than with his left. He never experienced any pain. Rubbing with his knuckles induced "wonderful stars" in his eyes as he argued. The Optical Coherence Tomography (OCT) of the cornea revealed no deposits. So, the cause of these changes looking similar to scars, is with high probability the constant intensive eye rubbing. Chronic micro traumata over 16 years may have led to those lesions. We informed him and his mother about these harmless side-effects. A corneal (lattice) dystrophy would look different and would be accompanied by a series of painful erosions.



Figures 1,2: Superficial white irregular shaped netlike crossing lines.

Reference

1. Stone EM (2007) Leber congenital amaurosis-a model for efficient genetic testing of heterogeneous disorders: LXIV Edward Jackson Memorial Lecture. Am J Ophthalmol 144: 791-811.