Different phenotypes of lattice corneal dystrophy type I in patients with 417c>T (R124C) and 1762A>G (H572R) mutations in Tgfbi (BIGH3)

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Purpose: To describe clinical data and to characterize mutations in the transforming growth factor beta-induced (TGFBI) gene in patients from three unrelated Chilean families with lattice corneal dystrophy type I (LCDI). Methods: Snellen acuity tests, anterior segment slit lamp examinations, dilated fundus evaluations, and tonometry were performed for seven patients-five females and two males belonging to three unrelated families-affected with lattice corneal dystrophy Type I. Genomic DNA was also extracted from peripheral leukocytes from the seven patients and four healthy relatives. The 417C>T mutation (R124C) was screened using PCR-RFLP for the seven patients and four healthy relatives. Exons 11, 12, 13, and 14 were sequenced in one patient not carrying the mutation in codon 124. Comparison of phenotype to genotype was performed. Results: The seven patients studied exhibited LCDI in both eyes, most of which were symmetric. Affected individuals demonstrated progression from central s