

Rapid Diagnosis of Most Common β igh3 Gene Associated Corneal Dystrophies by DNA Chip

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The aim of this study is to develop a rapid diagnostic DNA chip to detect mutations of most common β igh3 mutations which cause corneal dystrophies (CDs). Recent studies have shown that LASIK can worsen the CDs, and thus initial screening of β igh3 gene mutation is urgently needed before LASIK. Suitable primer and probe sets were designed to examine the exon 4 of the β igh3 gene in order to identify mutant and wild type alleles. DNA chip diagnosing these CDs was successfully developed. Mutations were then identified by signals of the probes immobilized on DNA chip developed in this study. Ninety eight participants volunteered for this test. Each DNA sample from peripheral blood was analyzed by DNA sequencing and DNA chip, and then compared with each result. We concluded that this rapid genotyping using DNA chip allowed successful detection of CDs with 100% sensitivity. We thus conclude that mutational analysis of exon 4 of the β igh3 gene can be successfully performed with DNA chip technology providing enough information for the diagnosis of CDs. [This work was supported by Medigenes Co., Ltd. and by the Brain Korea 21 Project. Further support through the LG Chemicals Chair Professorship is appreciated.]