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New findings in corneal dystrophies described from Korea Advanced Institute of Science and Technology

Biotech Week via NewsEdge Corporation :

2007 JUN 27 - (NewsRx.com) -- Fresh data on corneal dystrophies are presented in the report "Development of a DNA chip for the diagnosis of the most common corneal dystrophies caused by mutations in the betaigh3 gene." In this recently published study, investigators in Korea conducted a study "To develop a diagnostic DNA chip to detect mutations in the betaigh3 gene causing the most common corneal dystrophies (CDs). Samples from 98 people, including patients with betaigh3-associated CDs (beta-aCDs), were examined."

"Specific primer and probe sets were designed to examine exons 4 and 12 of the betaigh3 gene, in order to identify mutant and wild-type alleles. Mutations were then identified by hybridisation signals of sequence-specific probes immobilised on the slide glass. Direct sequencing of exons 4 and 12 of the betaigh3 gene in the patients' genome showed that beta-aCDs could be mainly classified into five types: homozygotic Avellino corneal dystrophy (ACD), heterozygotic ACD, heterozygotic lattice CD I, heterozygotic Reis-Bucklers CD and heterozygotic granular CD. Blind tests were performed by applying the target DNA amplified from the genomic DNA isolated from the peripheral blood of the participants onto a DNA chip. The results obtained by DNA chip hybridisation matched well with the direct DNA sequencing results. The DNA chip developed in this study allowed successful detection of beta-aCDs with a sensitivity of 100%. Mutational analysis of exons 4 and 12 of the betaigh3 gene, which are the mutational hot spots causing beta-aCDs, can be successfully performed with the DNA chip," wrote S.Y. Yoo and colleagues, Korea Advanced Institute of Science and Technology.

The researchers concluded: "Thus, this DNA chip-based method should allow a convenient, yet highly accurate, diagnosis of beta-aCDs, and can be further applied to diagnose other types of CDs."

Yoo and colleagues published their study in *British Journal of Ophthalmology* (Development of a DNA chip for the diagnosis of the most common corneal dystrophies caused by mutations in the betaigh3 gene. *British Journal of Ophthalmology*, 2007;91(6):722-7).

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