A Novel Decorin Gene Mutation in Congenital Hereditary Stromal Dystrophy: A Korean Family

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A 43-year-old man developed decreased vision in the right eye that had persisted for seven years. Under slit lamp examination, corneal clouding was noted with normal endothelium and ocular structure. From the clinical evidence, we suspected that the patient had congenital hereditary stromal dystrophy (CHSD). He and his family underwent a genetic analysis. Penetrating keratoplasty was conducted, and the corneal button was investigated for histopathologic confirmation via both light and electron microscopy. The histopathologic results revealed mildly loosened stromal structures, which exhibited an almost normal arrangement and differed slightly from the previous findings of CHSD cases. With regard to the genetic aspects, the patient and his mother harbored a novel point mutation of the decorin gene. This genetic mutation is also distinct from previously described deletion mutations of the decorin gene. This case involved delayed penetration of mild clinical symptoms with the histological feature of a loosened fiber arrangement in the corneal stroma. We concluded that this condition was a mild form of CHSD. However, from another perspective, this case could be considered as "decorin gene-associated corneal dystrophy," which is distinct from CHSD. Further evaluation will be required for appropriate clinical, histopathologic and genetic approaches for such cases.

Key Words: Decorin, Hereditary corneal dystrophy, Point mutation

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a flaky pattern of stroma was noted throughout the entire cornea. The right eye had decreased vision and exhibited relatively denser homogenous opacities than the left (Fig. 1).

The family members stated that corneal changes had been detected only in the patient’s mother at 69 years of age, and no specific issues had arisen in any other family member or relative. The patient’s father had reported no ophthalmic abnormalities before his death, and his mother had been diagnosed with diffuse corneal opacities of unknown etiology in both eyes three years previously (Fig. 2). She explained that she had experienced decreased vision since childhood, but these deficiencies produced no difficulties in her daily life. The patient’s brother and sister had no symptoms at all and no ophthalmic or systemic abnormalities. As far as the family knew, no one in the paternal or maternal lineage or offspring of the patient had experienced any eye problems except for the patient’s mother (Fig. 3).

The endothelium and Descemet’s membrane of the right eye were identified as normal following slit lamp examination. No gross abnormalities, such as Haab’s striae or features of posterior polymorphous corneal dystrophy, were detected in the right eye. The patient’s past medical records from another hospital demonstrated that his endothelial cells of both eyes presented with a normal shape and numbers under a specular microscope about six years ago. However, endothelial cells were found as indeterminate forms using specular microscopy due to the barrier of stromal opacity at the time of our study. The endothelial cells of the left eye were counted using a Konan Noncon

Fig. 1. Slit lamp photography of the patient. (A,B) Right eye. No gross abnormalities of the corneal endothelium, iris and lens were observed. Clouding of the cornea is noticeable under the arcuate slit beam. With magnification, ground-glass corneal opacities are more clearly seen in the anterior stroma, and identifiable small flakes and spots are present throughout the entire stoma. (C,D) Left eye. Density of corneal clouding is less than that of the right cornea.