

## **Subepithelial Amyloid Deposits in Congenital Hereditary Endothelial Dystrophy: A Histopathologic Study of Five Cases.**

### **Case Reports**

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#### **Abstract:**

**Purpose.** To report the clinical, histologic, ultrastructural, and immunohistochemical features of congenital hereditary endothelial dystrophy (CHED) associated with subepithelial amyloid deposits.

**Methods.** The clinical features of seven patients and histologic characteristics of eight corneal buttons were evaluated. The corneal specimens included five cases with histologic features of CHED associated with subepithelial amyloid. The remaining three corneal buttons of CHED without amyloid were obtained from the fellow eye of an affected patient and from siblings of two affected patients. Light microscopic studies were performed on sections stained with hematoxylin and eosin, periodic acid Schiff stain, and Congo red stain with and without permanganate bleach. Immunohistochemistry with an antibody to the amyloid AA protein and lambda and kappa light chains was done on all specimens. Electron microscopy was performed on three corneal specimens. The cases were followed for 1-9 years.

**Results.** The notable clinical findings included decreased vision, history of parental consanguinity (4/7 cases), and affected siblings (5/7 cases). Examination revealed nystagmus (5/7 cases) and bilateral ground-glass corneas in all patients. In addition, central subepithelial whitish opacities were noted in patients with CHED and amyloid. Three patients had associated congenital glaucoma. The patients underwent penetrating keratoplasty at a mean age of 10 years. Histologically, five corneal buttons of CHED revealed varying degrees of subepithelial amyloid deposits associated with a subepithelial fibrous pannus. Immunohistochemically, the deposits were nonreactive to anti-amyloid A antibody but were immunoreactive with an antibody to lambda light chains in two cases. Electron microscopy confirmed the presence of subepithelial amyloid. Thickening of Descemet's membrane and attenuation of corneal endothelial cells, noted in all cases, was consistent with features of CHED. The corneal buttons from the fellow eye and the siblings showed histologic features of CHED,

with a subepithelial fibrous pannus without amyloid deposits. Spheroidal degeneration was noted in two corneal specimens. To date, no recurrence of the amyloid deposits has been seen in the grafts.

**Conclusions.** This study demonstrates that subepithelial amyloidosis may be rarely associated with a recessive form of congenital hereditary endothelial dystrophy. The clinical, histologic, and immunohistochemical features suggest a secondary form of amyloidosis.