Abstract glaucoma

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Primary congenital and developmental glaucomas.

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Abstract

Glaucoma is the leading cause of irreversible blindness worldwide. Although most glaucoma patients are elderly, congenital glaucomaand glaucomas of childhood are also important causes of visual disability. Primary congenital glaucoma (PCG) is isolated, non-syndromic glaucoma that occurs in the first three years of life and is a major cause of childhood blindness. Other early-onset glaucomas may arise secondary to developmental abnormalities, such as glaucomas that occur with aniridia or as part of Axenfeld-Rieger syndrome. Congenital and childhood glaucomas have strong genetic bases and disease-causing mutations have been discovered in several genes. Mutations in three genes (CYP1B1, LTBP2, TEK) have been reported in PCG patients. Axenfeld-Rieger syndrome is caused by mutations in PITX2 or FOXC1 and aniridia is caused by PAX6 mutations. This review discusses the roles of these genes in primary congenital glaucoma and glaucomas of childhood.