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PRIMARY CONGENITAL GLAUCOMA – MOLECULAR MECHANISMS – GENETICS.

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Introduction

Primary congenital glaucoma (GCP) is an eye condition caused by the abnormal development of aqueous humor drainage structures, characterized by increased intraocular pressure, enlargement of the eyeball, corneal edema and changes of the optic nerve.

Keywords

genes, primary congenital glaucoma, chromosome , mutation.

Purpose

Locus	Location	Inheritance	Mutated gene (MIM number)
GLC3A	2p21	AR	*CYP1B1* (601771)
GLC3B	1p36	AR	Unknown

AR: Autosomal recessive, MIM: Mendelian inheritance in man

Material and methods

This study was based on the synthesis and analysis of literature from open access databases: Pubmed, Scopus; GoogleSchoolar, Hinari.

Results

Genetic mapping of affected gene families has identified several chromosomal loci that cause primary congenital glaucoma: GLC3A (chromosome 2p22), GLC3B (chromosome 1p36.2 – p36.1), GLC3C (chromosome 14q24.3), GLC3D (chromosome 14q24.2-q24.3) and GLC3E (chromosome 9p21.2). Mutations have also been identified in the LTBP2 (14q24.3) genes encoding the latent – transforming growth factor 2 beta – binding and MYOC (14q23 – q24) encoding the myocillin protein for role in cytoskeleton organization and cell adhesion, TEK (tyrosine kinase receptor), COL1A1. Mutations in the CYP1B1 gene (missens, insertions and/or,del) encoding the P450 protein with a role in the metabolism of endogenous molecules necessary for ocular development leading to autosomal recessive GCP have been shown to be a strong risk factor.

Conclusions

Primary congenital glaucoma is a genetic disease caused by mutations in different genes (GLC3A, GLC3B, GLC3C, GLC3D, GLC3E, LTBP2, MYOC, TEK, COL1A1, CYP1B1) and population screening through genetic testing can reduce the incidence of the disease and can be helpful to clinicians for a personalized approach to treatment.