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The Genetics and the Genomics of Primary Congenital Glaucoma.

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Abstract

The sight is one of the five senses allowing an autonomous and high-quality life, so that alterations of any ocular component may result in several clinical phenotypes (from conjunctivitis to severe vision loss and irreversible blindness). Most parts of clinical phenotypes have been significantly associated with mutations in genes regulating the normal formation and maturation of the anterior segments of the eye. Among the eye anterior segment disorders, special attention is given to Glaucoma as it represents one of the major causes of bilateral blindness in the world, with an onset due to Mendelian or multifactorial genetic-causative traits. This review will point out the attention on the Primary Congenital Glaucoma (PCG), which is usually transmitted according to an autosomal-recessive inheritance pattern. Taking into consideration the genetic component of the PCG, it is possible to observe a strong heterogeneity concerning the disease-associated loci (GLC3), penetrance defects, and expressivity of the disease. Given the strong PGC heterogeneity, pre- and posttest genetic counseling plays an essential role in the achievement of an appropriate management of PCG, in terms of medical, social, and psychological impact of the disease.