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Meta-analysis of CYP1B1 gene mutations in primary congenital glaucoma patients

Amine Haddad
University of Casablanca, Morocco

Glaucoma affects more than 70 million people worldwide, which making it the second leading cause of blindness in the world.1 The rare and severe form of glaucoma called, Primary congenital glaucoma (PCG), is usually transmitted as an autosomal-recessive disease with an incomplete penetrance and about 10% to 40% of its cases are familial.2,3 The importance of this presentation is to highlight the interest of the genetic study of CYP1B1 mutations, its impact on the development of the eye and the genesis of PCG. This information could help clinicians to identify the population at high risk, to screen for genetic tests and to act early in a way to improve the patient prognosis. An in-depth study was carried out by the following search engines: PubMed, Scopus, clinic key and direct science for articles that have been published from 2011 until 2020. We have identified 81 articles in this review from 2011 to 2020, excluding 3 duplicates. 78 titles and abstracts were filtered from which 52 studies were not considered since they did not match the inclusion criteria as mentioned above. 26 complete studies were examined. One hundred and sixty-one mutations were found in 1641 tested patients and three families, including 78 novel mutations. We identified a no significant difference in the sex ratio and the bilaterality was reported in the majority of patients. We have shown through this study that inbreeding plays an important role in the pathogenesis of PCG transmission compared to the sporadic mutations that have been found in some cases. The majority of the included studies were from ASIA (64.3%), followed by Europe (17.85%), America (10.71%) and Africa (7.14%). The first and most common mutation in our study is 182 G>A (p.Gly61Glu). Considering the precedent analysis, our study encompasses a higher number, for a better understanding of the association of these mutations with PCG. Therefore, it can be useful to develop reliable genetic tests to be a routine examination for patients with a family history of PCG.

Biography

My name is Amine Haddad, I am in the 4th year of my specialty in ophthalmology in Morocco and I am currently doing an internship in Paris. I have always been fascinated by the genetics of ophthalmological diseases, which is why I chose the genetics of glaucoma as the subject of my PhD in the Laboratory of Medical Genetics and Molecular Pathology, Department of Medicine and Pharmacy, Hassan II University of Casablanca, Morocco.