

# MOLECULAR STUDIES AND CLINICAL EVALUATION OF PRIMARY CONGENITAL GLAUCOMA

## ABSTRACT

**PURPOSE :** Clinical evaluation of primary congenital glaucoma and Identification of polymorphism in *CYP1B1* gene in patients diagnosed to have PCG.

**METHODOLOGY:** 20 patients of Primary Congenital Glaucoma are registered for this study conducted in the Regional Institute of Ophthalmology and Government Ophthalmic Hospital, Chennai. Detailed clinical evaluation was done including measurement of corneal diameters, measurement of intraocular pressure and fundus examination. Blood samples will be collected from patients after informed consent. DNA isolated from these samples will be subjected to PCR for amplifying the coding exons of the *CYP1B1* gene using gene-specific oligonucleotide primers. PCR products will be sequenced and analyzed to identify mutations in *CYP1B1*.

**RESULTS:** Out of the 20 children who were diagnosed to have Primary Congenital Glaucoma before the age of 1, 10 % of the children presented with symptoms of PCG within 1 month after birth, 65 % of them presented within 1 to 6 months of age and 25 % of them presented after 6 months. 90% of the children had bilateral involvement. Only 5 % of the eyes had clear cornea. 62.5 % of eyes had mild haze where fundus could still be visualized. 32.5% had severe haziness where it was not possible to visualize the fundus. Common Single Nucleotide Polymorphisms which are suggestive of published *CYP1B1* mutations in PCG individuals are found in 2 patients out of the 20 patients included in this study group.

## **CONCLUSION:**

The technological advances in molecular diagnostics and genetics can aid us to characterize pathologic gene changes providing further insight into the molecular pathogenesis of this disorder and improve management strategies of PCG including genetic counselling.

**Key words:** CYP 1B1 mutation, Primary Congenital Glaucoma, single nucleotide polymorphisms