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Analysis of CYP1B1 gene in sporadic primary congenital glaucoma affected cases from Pakistan

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Statement of the Problem: Primary congenital glaucoma (PCG) is primary cause of blindness in early age. Mutations in CYP1B1 gene are frequently associated with PCG especially in inbred populations. The aim of this study was to identify the genetic mutations in CYP1B1 gene in sporadic PCG cases belonging to 17 consanguineous families from Pakistani population.

Methodology: 17 consanguineous Pakistani families segregating autosomal recessive PCG were enrolled. Genomic DNA was extracted and amplification was done by using primers flanking coding exons of CYP1B1 using genomic DNA from affected and selected unaffected individuals. Novel variant was analyzed in 96 ethically matched controls from Pakistani population with no history of glaucoma.

Findings: The sequence analysis results revealed a novel splice site variant and three already reported missense mutations p.G61E, p.R390H and p.E229K in CYP1B1 gene, which segregates with the disease in respective families. In addition two missense variations p.N453S and p.R48G were also found in two patients segregating with disease phenotype. Novel splice site variant was not detected in controls.

Conclusion & Significance: Six (6) out of seventeen (17) children with PCG (35%) had disease due to homozygous CYP1B1 alterations suggesting that CYP1B1 is a major gene contributing to PCG in Pakistani sporadic patients. Identification of a novel mutation indicates genetic heterogeneity of disease. Our findings will help to provide genetic counseling to the affected families to minimize new suffering individuals in these families.

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