Chikezie Grand Ihesiulor et al., Optom open access 2018, Volume 3
DOI: 10.4172/2476-2075-C1-008

Molecular basis of congenital glaucoma

Chikezie Grand Ihesiulor[1] and **Udo A Ubani**[2]
[1]The University of Manchester, UK
[2]Abia State University, Nigeria

Purpose: The purpose of this study was to detect pathogenic mutations in cytochrome P450, family 1, subfamily B, polypeptide 1 *(CYP1B1)* gene in 19 sporadic primary congenital glaucoma (PCG) cases and to identify patients lacking *CYP1B1* mutations. Secondly, to conduct an *in silico* analysis of exome sequencing data of variants common to three related pigment dispersion syndrome (PDS) patients.

Methods: *CYP1B1* exon 2 and the coding part of exon 3 of 15 participants were amplified by polymerase chain reaction and amplicons were sequenced by Sanger sequencing. Sequencing data was analyzed to identify the gene mutations or SNPs. Second, the exome sequencing data of the PDS patients combined was analyzed in-house by bioinformaticians and further filtered manually to identify candidate genes for PDS.

Results: Four previously reported PCG-associated *CYP1B1* mutations (c.1159G>A; p.E387K, c.230T>C; p.L77P, c.1103G>A; p.R368H and c.1568G>A; p.R523K) were found in four patients out of the 15 fully 'sequenced' patients. Also, 10 previously reported single nucleotide polymorphisms and two novel noncoding variants were identified. Second, 21 candidate genes were found after filtering using various databases (OMIM & GeneDistiller). Nine genes (*TPCN2, TYR, PAX6, DICER 1, FOXE3, TGIF1, TCF4, RPGR and CNGB3*) may be of more importance since they are associated with ocular diseases.

Conclusion: The relatively low percentage of PCG patients having CYP1B1 mutations (4/15=26.6%) demonstrates that other known and unknown genes may contribute to PCG pathogenesis. Lack of CYP1B1 gene mutations in some patients stresses the need to identify other responsible candidates. More analysis may be needed and the genes identified may be screened in future in other PDS patients to study PDS genetics.

Biography

Chikezie Grand Ihesiulor was born in Port Harcourt, Nigeria, in 1986. He received the O.D. Doctor of Optometry degree in Optometry from Abia State University, Uturu, Nigeria, in 2008 and the MSc in Investigative Ophthalmology and Vision Sciences in The University of Manchester, UK in 2013. In 2015, I joined the Department of Optometry, Abia State University, as a Lecturer. His current research interests include glaucoma, ocular trauma, ocular genetics, preventive optometry, visual psychology and psychopathology. Dr. Chikezie is a member of the Nigerian Optometric Association (NOA) and the Optometrist and Dispensing Opticians Registration Board of Nigeria. He is the CEO of Healthy-hope Lifestyle Centre, Nigeria. He was awarded the best clinician by the President, NOA in 2009 and has joined and led several community health care teams in Nigeria and UK to offer free medical and eye health care services. He is an innovative researcher and is currently pursuing his Ph.D. in Optometry in The University of Manchester, UK.

Notes: