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EDITORIAL

Influence of genetic susceptibility in visual impairment

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Editorial

Visual impairment is a significant decrease or loss of vision which is considered as an important public health concern. The prevalence of visual impairment is more frequent in developing countries as compared to the developed nations. It can be successfully corrected by early diagnosis and treatment [1,2]. Various factors associated with visual impairment includes glaucoma, uncorrected refractive errors, cataract, corneal opacity, age-related macular degeneration, quality of health service, vitamin A deficiency, sex, age, school type, duration and distance of television exposure, mobile use and the socioeconomic status [3,4]. According to World Health Organization (WHO) estimates 253 million individuals sustain vision impairment, of which 217 million have moderate-to-severe vision impairment and 36 million are blind [3]. Over 80% of all visualdisorders can be prevented or cured [3]. In developing countries, this frequency could be as high as 90% mainly affecting individuals older than 50 years [5,6]. In children of school-age, visual impairment may have a drastic impact on educational performance. It has been estimated that 75%–90% of all learning in the classroom has been attributed to either wholly or partially via the visual pathway [7]. This results in a negative effect on learning and social interaction, thereby affecting the natural development of academic and social abilities [7,8].

Genetic susceptibility plays a crucial role in various types of visual impairments that might result in blindness in infants, pediatrics and adults [9]. Currently, molecular diagnosis of genetic eye diseases is considered to play a major role in improving clinical management. Of note, clinically similar or dissimilar diseases evolved due to phenotypic heterogeneity caused by mutations [10]. Therefore, detection of the underlying genetic mutations plays a vital role in confirming the clinical diagnosis, prognosis and therapeutic management. For instance, Cerebral visual impairment (CVI) is a primary cause of visual dysfunction in pediatrics due to inappropriate projection and/or interpretation of the visual input to the brain. The acquired causes of CVI are well established; still there exist a need to explore the novel

genetic factors associated with underlying CVI. Bosch DG et al [11] performed the whole-exome sequencing in 25 patients with CVI and intellectual disability. The authors suggested the involvement of more than one candidate genes for the genetic diagnosis for CVI. Notably, a novel PGAP1 variant has been identified to have genetic association with CVI. It has been suggested that the whole exome sequencing is an appropriate diagnostic tool for the detection of de novo mutations which confirms the genetic diagnosis of intellectual disability [12]. Genetic eye disease (GED) is another cause of blindness which includes disorders involving all structures of the eye. Therefore, molecular diagnostics is promising in identifying genotype-specific novel targets and treatments to utilize the current knowledge of genetic susceptibility with complex ocular disorders [13].The findings provide an overview and insight to genetic causes underlie CVI [11].

Early diagnosis and adequate treatment of visual disorder require development of appropriate genetic diagnosis tools and cost-effective interventions to reduce the burden of disease related to loss of vision in the developing nations with disproportionate socio-economic status. The genetics of inherited vision disorder is a promising area of research that needs further attention and focus in developing nations.

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