DBT-NIBMG scientists gain deeper molecular insight on Glaucoma

New Delhi, June 09: Glaucoma is one of the leading causes of irreversible blindness worldwide. It is primarily caused due to an increased pressure in the eye called intraocular pressure (IOP) resulting in damage to the optic nerve.

Primary angle closure glaucoma (PACG) is a major subtype of the disease, prevalent mostly in east and south Asia including India and China. A narrow angle in the front part of the eye, between the iris and the cornea, is considered as the primary risk factor for this subtype. The narrow angle could be responsible for high IOP and subsequent degeneration of retinal ganglion cells. However, only 50% individuals with complete angle closure progress to develop glaucoma. There are several underlying genetic causes. But they are complex in nature and molecular mechanisms remain elusive.

A team of researchers in the Department of Biotechnology's National Institute of Biomedical Genomics led by Dr Moulinath Acharya in association with a group of scientist in All India Institute of Medical Sciences, New Delhi conducted a study using young onset cases of the disease who are likely to be enriched in genetic susceptibility factors and compared them

with older controls with a narrow angle but no glaucoma; these older individuals are likely lacking genetic factors responsible for PACG. The aim was to identify the genes responsible for development of glaucomatous neurodegeneration in individuals with enriched genetic susceptibility.

Through their work that was based on genome-wide association analyses, Dr. Acharya and his colleagues identified genetic variations in a gene called *ABCA4*, which is found in the light sensitive cells in the retina. They also found genetic variations in genes involved in relevant biological pathways that could potentially be involved in IOP variation and glaucoma pathogenesis.

They also identified another gene called *CNTNAP5*. This gene is involved in cell adhesion and intercellular communication in neuronal cells. It is expressed in the brain and it interacts with other associated genes found in the study, thus making it an interesting candidate for future functional investigation.

Overall, the results indicated a strong genetic association of *CNTNAP5* locus with PACG and suggest its potential involvement in glaucomatous neurodegeneration, an essential component of PACG pathogenesis. Additionally, pathways relevant to cell adhesion and cell death may be important in disease pathogenesis.

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