Sankara Nethralaya zeroes in on genes causing glaucoma

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CHENNAI: Scientists at Sankara Nethralaya are part of an international group that has discovered three genetic locations associated with a form of glaucoma common in the Asian population.

The study, which has been ongoing for the last 10 years, sought to find explanations for why only some people with the susceptibility go on to develop the disease. The results were published in a recent issue of *Nature Genetics*. Primary Angle Closure Glaucoma (PACG) is common in people of Indian and Chinese descent. An estimated 15 million people afflicted with PACG live in Asia. It is also said to be responsible for a substantial proportion of blindness in many Asian countries.

The team from Sankara Nethralaya Vision Research Foundation, headed by L.Vijaya, who also heads the glaucoma division, and comprising Ronnie George, Sripriya Sarangapani and Nagaswamy Soumittra, worked with the international team to narrow in on the specific gene mutations responsible for PACG.

GLAUCOMA-TRACKING THE GENES

NORMAL VISION | VISION WITH GLAUCOMA

This form of glaucoma arises from high pressure in the eye, hindering drainage from it.

"There is a pre-disease phase in glaucoma, in PACG, where the condition is remediable. But what we have also seen is that not everyone who has pre-disease goes on to develop glaucoma," explains Dr. Ronnie George, senior consultant, Glaucoma, and research director. Only a percentage of this suspect group develops glaucoma. "In India, there are about 24 million people above the age of 40 who are susceptible. But it will be unnecessary to begin blanket treatment for all."

Therefore, research began to identify the risk factors which predict who will progress on to glaucoma. "PACG runs in families, so we began looking at families to identify gene mutations accounting for the disease. But that was not successful, because multiple genes were involved and this is a complex disease."

"The best thing is a genome-wide analysis. When the Human Genome Project mapped all the genes, it provided markers for the genes (in the single-nucleotide polymorphism). These are like milestones on the road. We also know which genes are close to which milestone, so by tracking the milestone/marker, we get at the gene," explains Dr. George.

Researchers then compared the milestones among patients with the disease, and those who are normal to identify those that are different, to zero in on these three genetic locations. It is likely that these are associated with an increased risk for the disease."

"In order to confirm, we tested for the same in a second group of patients, also with the disease. Only if we see it in both groups can we say for sure that there is a link," according to Dr. George. These however, account only for about 10 per cent of the disease.

It is necessary to identify at least 15 gene mutations that will hold for all cases of PACG, he adds.