

MedGenome and Narayana Nethralaya Organised a Discussion on Genetics in Ophthalmology

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Genetic Testing is an Effective Tool to Screen Ocular Cancers

MedGenome and Narayana Nethralaya, Bangalore organised a discussion on '**the relevance of genetics in the clinical practice of ophthalmology**'. Eminent practitioners as Dr. Arkasubhra Ghosh, Head of Department, Grow Research Laboratory, Narayana Nethralaya; Dr Anuprita Ghosh, Scientist and Genetic Counsellor, Grow Research Laboratory, Narayana Nethralaya; Dr Himika G and Dr Fairooz Manjandavida ophthalmologists at Narayana Nethralaya participated in the discussion. The doctors opined that certain ocular conditions like retinal dystrophies or ocular cancers like retinoblastoma can be screened for by genetic testing to help manage the condition, and the advances in genetics will hopefully provide us a cure for these conditions in the future.

The genetic basis of eye diseases is understood much better now than before. Retinal Dystrophy, the name given to a range of ocular conditions affecting the retina, associated with reduced or deteriorating vision in both eyes, is more than not, a genetic disease. Retinitis pigmentosa (RP) is the most common inherited form of retinal degeneration, affecting nearly 1/4,000 individuals. Leber Congenital Amourosis (LCA) is another retinal disorder that results in visual impairment starting during early childhood. The gene *RPE65* is one of the already known genes that has been implicated in LCA. Gene replacement therapy trials for genetic retinal disorders in animals have been found successful. Human clinical trials are on-going which provides hope to the clinicians about a possible curative treatment in the future.

Retinoblastoma, a rare type of eye cancer that usually develops in early childhood, occurred at the rate of 3-5 per million children per year, and accounts for 2.5 to 4% of all childhood cancers in most developed countries. One out of three children with retinoblastoma develops cancer in both eyes. Retinoblastoma is caused by a mutation in the RB1 gene. When diagnosed early, it can be removed before it spreads beyond the eye to other parts of the body. If left untreated, this advanced form of retinoblastoma can be life threatening.

The clinicians discussed genetic testing as an effective tool for familial screening in both the above conditions, to identify proactively the other family members who might be afflicted with the disease, and act as a guide for further family planning measures.

The implicated genes for the aforementioned eye diseases have been identified and studied, thereby making genetic testing for their diagnosis a very viable option. Narayana Nethralaya provides free retinoblastoma screening to needy patients in collaboration with MedGenome, a state-of-the-art next-generation sequencing laboratory co-located at the Nethralaya premises. Over 75 patients have so far availed of this opportunity where the Narayana Nethralaya doctors support the patients from pre-counselling through treatment up until post-treatment counselling.

The discussions also revolved around the future of gene therapy for ocular disease treatments. The eye is an ideal target for gene therapy. Relatively small and highly compartmentalized, eye is an immune privileged organ with well-defined targetable diseases known to benefit from prolonged therapy. It is also fairly easy to distinguish both potential side effects and treatment benefits. The clinicians were in agreement that the future of ophthalmology lies in molecular diagnostics and gene therapy and initiatives to increase the awareness about these advances among the medical community is a need of the hour.