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MedGenome & Narayana Nethralaya deliberate on need for genetic tests to detect ophthalmology diseases

Our Bureau, Bengaluru, **Thursday, March 03, 2016, 14:05 Hrs [IST]**

MedGenome and Narayana Nethralaya have indicated that certain ocular conditions like retinal dystrophies or ocular cancers like retinoblastoma can be screened for by genetic testing to help manage the condition.

The advances in genetics will hopefully provide us a cure for these conditions in the future, stated scientists and clinicians in a discussion on the relevance of genetics in the clinical practice of ophthalmology.

Dr. Arkasubhra Ghosh, head of department, Grow Research Laboratory, Narayana Nethralaya, Dr Anuprita Ghosh, scientist and genetic counselor, Grow Research Laboratory, Narayana Nethralaya, Dr Himika G, and Dr Fairooz Manjandavida ophthalmologists, Narayana Nethralaya participated in the discussion which also revolved around the future of gene therapy for ocular disease treatments.

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 is 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 ongoing which provides hope to the clinicians about a possible curative treatment in the future, they said.

The eye care major provides free retinoblastoma screening to needy patients in collaboration with MedGenome, an advanced next generation sequencing laboratory. Over 75 patients have so far availed of this opportunity where the doctors support the patients from pre-counselling through treatment up until post-treatment counselling.

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 per cent 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, pointed out the team of experts.

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.

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