Genetic Testing for Eye Diseases

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Advances in ocular genetic research allows a better understanding of the genetic basis of many eye diseases. Genetic testing is becoming available for more and more disorders and the number of genes being discovered is also on the rise, although the testing may not be accessible to all because of the costs involved.

Finding the genetic basis for a patient’s disease allows for risk prediction, accurate diagnosis, focused treatment and preventive screening and care. Personalized medicine, particularly in the field of pharmacogenetics, is becoming a reality and we may be able to offer treatments to patients based on the genetic testing results. We do know that some patients respond and some do not to the same anti-VEGF treatment for AMD/PCV and part of this could be because of the genetic makeup, therefore if we have the results with us before the start of the treatment, we may be able to decide better for the patient. However, genetic testing for AMD is still in its infancy.

With better understanding of retinal degenerations, we now know that patients with ABCA4 mutations should not be given Vitamin A prophylaxis as it can worsen their disease. So with this knowledge the older practice of putting RP patients on vitamin A prophylaxis has reduced. Whilst there is no effective therapy that we can offer to these patients, we should not cause a further decline in their visual status.

Gene therapy trials have taken place for RPE65 gene and for choroderemia and more trials are in early phases.

Genotype-phenotype correlation for a few conditions have made us understand the diseases much better. Knowing that a child with aniridia has WAGR syndrome, we can be more vigilant in systemic work up.

Importance of genetic testing for Retinoblastoma cannot be overemphasized. Children with germline mutation are at risk for developing second non-ocular cancers and can also pass the defective mutation to the offspring and this could be explained to the family. For non-germline cases we can avoid unnecessary anesthesia exams. This can also be a big cost saving for the family and can avoid unnecessary long travels to specialized centers.

At Sankara Nethralaya, we have now started offering genetic testing for a lot of eye conditions after a pedigree-based counseling and the results are then handed over to the patient after proper verification. The whole process takes around 4–6 months.

I would recommend all readers to read the article by Ed Stone et al. in Ophthalmology about recommendations for genetic testing of inherited eye diseases.

Reference

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