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Editorial

Ocular Genetics: A Sub-specialty Service for Genetic Eye Diseases

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Genetic eye disease (GED) is one of the leading causes of blindness and includes disorders affecting all structures of the eye from the anterior to posterior segment, such as albinism, corneal dystrophy, aniridia, primary glaucoma, pediatric cataracts, retinitis pigmentosa, Stargardt disease, and hereditary optic neuropathy.[1] Many systemic genetic diseases and syndromes such as Marfan syndrome, neurofibromatosis, mitochondrial disorders, and chromosomal abnormality syndromes have significant ocular pathology. Whilst rare individually, together these disorders are a significant cause of blindness and visual impairment. They are particularly important in children and confer a significant burden in the working population. It is estimated that each year around 150 children and 250 adults of working age are newly diagnosed as blind or partially sighted, as a result of a genetic disorder.[2] In Oman, visual disability due to nutritional and communicable eye diseases has declined and the proportion of blindness due to diseases of posterior segment of the eye (diabetic retinopathy); however, glaucoma and genetic disorders have increased.[3] Childhood blindness remains a challenge with hereditary disorders accounting for severe visual impairment/blindness in a third of all children.[4]

Only 60-70 years ago in 1943, Oswald Avery proved that deoxyribonucleic acid (DNA) carries genetic information, and in 1953, Watson and Crick discovered the structure of DNA. Since then, the field of genetics has been transformed. The Human Genome Project with the complete sequencing of the human genome,[5] and the Hap Map Project with its unprecedented view of single nucleotide polymorphism (SNP) variation,[6] have induced enormous changes in the human genetic landscape, changing the way in which we view diseases and receive medical care. Now, with the aid of powerful new technologies for the manipulation and analysis of DNA, scientists are able to locate and identify genes responsible for synthesizing proteins, characterize their mutations and gain a deeper understanding of many poorly understood or previously unknown causes of disease.

In ophthalmology, the number of genes known to cause Mendelian genetic disease has greatly increased over the past decade. Researchers have discovered numerous mutations that lead to disease. This new knowledge has been shown to be crucial in the management of GED and has resulted in a paradigm shift in diagnosis and therapy.[7,8] Although some of these advancements may be well-known, it may not be possible for the general ophthalmologist to keep abreast with all the developments in the field of ocular genetics. Individually, GEDs are rare and general ophthalmologists are likely to have little experience of diagnosis and counseling. Increasing understanding of the importance of comprehensive care encompassing diagnosis, therapy, rehabilitation, and counseling for such patients, and for advancements in genetics to positively impact the way ophthalmology is clinically practiced, ocular genetics has emerged as a subspecialty in Ophthalmology.

A validated approach to genetic diagnosis in an ocular genetics service commences with elaborate history-taking (of the patient and family) and creation of a family pedigree, followed by careful clinical examination (affected people as well as those apparently unaffected); and lastly, clinical and genetic testing.[9] By their nature, genetic disorders may affect other family members. The ocular genetics service is able to encompass the whole family rather than solely the index individual referred. Clear communication is a defining issue of good service. Patients want information on their diagnosis delivered in a manner that they can understand. Genetic counseling which combines the provision of risk information with a support function is an integral part of any ocular genetics service. Patients and their families highlight the value of genetic counseling but report that it is often not offered in eye clinics.[10] Patients note that only when the person has psychologically adjusted to the condition can he or she move forward and start to deal with the implications. Many patients comment on the shock they experience on being given a diagnosis and that no support is offered to help them adjust to the information and its implications.

Ocular genetics enhances mutual understanding and communication between ophthalmologists, clinical and molecular geneticists, as well as genetic counselors. Many patients with GED have complex needs and their sight problems may be one of a range of symptoms they experience as a result of their genetic condition. Comprehensive ocular genetics services where ophthalmologists work alongside clinical geneticists in joint clinics or liaise closely with the genetics services ensure that families receive a high quality and comprehensive package of care.

Ophthalmology is an area of mainstream medicine where molecular genetic testing is becoming an important aspect of the service. Genetic testing in ophthalmology has been demonstrated

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to have clinical utility through increased information from better diagnosis and prognosis, through decreased morbidity and mortality from preventive care and informed treatment options, and from provision of information to assist reproductive choice. Molecular diagnosis will become increasingly important with the development of novel treatments that are genotype specific, and as our knowledge of genes associated with susceptibility to complex disorders increases. An ocular genetics service offers patients to be part of a research effort. Patients and families understand that helping research will lead to more accurate diagnosis and therapies for themselves and their families. Patients also want to be kept informed of advances in research. All these aspects of care require more time than is available in routine ophthalmology clinics, and is well taken care of in the ocular genetics services.

Health systems need to evaluate and prioritize new scientific and medical knowledge and technologies, and integrate them effectively into their service and practice. To give patients with GED and their families the full benefit of expanding genetic knowledge, ocular genetics must be recognized as an independent and necessary sub-specialty discipline in Ophthalmology.

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