Ocular genetic abnormalities occur due to alterations in DNA. Such changes might affect various cellular structures and functions of the eye contributing to an array of ocular disorders, which may or may not be reversible. These genomic changes could be spontaneously occurring or inherited as a result of a pre-existing mutation.

In 2010, a global estimate showed that 285 million people were visually impaired, of which 39 million were blind and 246 million had low vision. Those visually impaired (about 35%) ranged from infancy to 49 years of age and the major causative factors were of a non-communicable and undetermined nature.

Non-communicable diseases (NCD) are increasing, and ocular related disorders are not exempt from this group. The rise in ocular diseases requires attention considering the negative impact it will have on the global economy, especially in developing nations.

In Oman, the causes of visual impairment are changing from communicable treatable diseases to incurable chronic ones with lifelong consequences. Although improved health services and initiatives have assisted in lowering some of the visual impairment cases, further intervention is needed as the transition to NCD continues.

Environmental factors (e.g. ultraviolet radiation) play a role in ocular disease manifestation, but there are genetic components associated with NCD. This becomes an issue of personal, societal, and governmental concern when disease-causing alleles are at high frequencies in the populations. Cultural preferences, such as consanguineous marriages (56.4% of all unions in Oman), endogamous unions, and bearing of many children, might increase allele frequencies in a population. As these practices are common in Middle Eastern countries the chances of individuals inheriting a recessive disease trait and possibly several ancestral disease alleles is elevated. Homozygosity or autozygosity mapping is a suitable method for disease gene detection in most cases. It can be used for the identification of known or novel variations in autosomal recessive conditions, even if a singleton sample pool is available. An example of this is the detection of a new variant in the FAM161A gene of a Palestinian family causing retinitis pigmentosa. Homozygosity mapping in combination with exome sequencing is also an option. Although exome sequencing has limitations, such as the difficulty in distinguishing disease from non-disease causing variants, it compensates for limitations associated with homozygosity mapping, where dominant, X-linked and compound heterozygous mutations cannot be detected. Both approaches have been used to identify a novel autosomal recessive retinitis pigmentosa variant with late-onset hearing loss in a Middle Eastern family.

Considering the cultural practices, genetic research as well as epidemiological investigations becomes of immense importance for Oman. The results obtained would enhance the understanding of the underlying disease pathophysiology and would facilitate drug-design and the development of treatments and therapeutic strategies. So, where does Oman stand with regard to ocular genetic research?

Approximately 313 publications relating to ophthalmology have been associated with Oman. These were found in the PubMed database using ophthalmology and Oman as keywords. Publications retrieval was set from the year 1980, before the construction of the first university in Oman, to the present date. Only two articles on the genetics of ocular diseases existed. Without a doubt all research fields are important, but the
low number of publications from Oman on ocular genetic research is of concern. The acquisition of such information is essential in determining the role of these genes in the population. Additionally, the initiation of translational research or targeted gene therapy requires basic knowledge of the genetic disease, such as gene location, altering mutations, and functional effect. Furthermore, the concept of private mutations, globally encountered, probably occurs in the Omani population too. However, with limited ocular genetic research, the identification of mutation profile of ocular genetic variants in Oman and their implications is limited.

Ocular genetic disorders have been the center of gene therapy success stories, such as the restoration of sight in adults and children with retinoblastoma. Patients received injections of an adenoviral vector containing a herpes simplex thymidine kinase gene with no systemic immune response. In patients with Leber congenital amaurosis administration of a recombinant adeno-associated virus transporting the retinal pigment epithelium-specific protein 65kDa gene resulted in improved retinal and visual function measurements. Surprisingly, sometimes gene therapy may be self-mediated. An example is the spontaneous chromothriptic cure of an individual with a rare congenital immunodeficiency disorder known as warts, hypogammaglobulinemia, infections, and myelokathexis syndrome (WHIM), which is caused by mutations in the chemokine receptor CXCR4 gene. However, cellular events in which chromosomes undergo massive deletion and rearrangement resulted in the splicing out of the mutated gene in a hematopoietic stem cell. This cell then repopulated and restored the immune function.

Ocular genetic research is fascinating not only in terms of therapeutic outcome, but also in the extraordinary cases of vision perception. An example of enhanced vision is the ability to perceive color spectrums unnoticed by the ordinary person (behavioral tetrachromacy), color spectrums unnoticed by the ordinary person example of enhanced vision is the ability to perceive the extraordinary cases of vision perception. An

**REFERENCES**