Michael Dean, J Clin Exp Ophthalmol 2014, 4:5
http://dx.doi.org/10.4172/2155-9570.S1.017

OMICS Group Conferences
Accelerating Scientific Discovery

4th International Conference on
Clinical & Experimental Ophthalmology
July 14-16, 2014 DoubleTree by Hilton Baltimore-BWI Airport, USA

Insights into eye diseases and biology using genetics

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The eye has represented the organ most accessible to clinical observation and one of the organs for which patients regularly seek treatment for dysfunction. As such, the genetics and biology of eye diseases have a rich history. In addition, because of the complexity of the visual cycle and architecture of vision, there are numerous inherited and complex disorders affecting vision. Examples will be provided of dominant, recessive, and X-linked ophthalmic diseases that have been analyzed and understood using molecular genetics and medicine. Furthermore, an update on the complex disease, age-related macular degeneration (AMD), the most common cause of vision loss will be presented. AMD is one of the more complex diseases that is best understood by genetic analysis, with examples of common variants with moderate effect (i.e. in genes CFH, CFH, ARMS2) as well as rare variants with strong effect (i.e. in genes ABCA4, CFH). These examples provide an informative context to understand recent progress on Schnyder Corneal Dystrophy.

Biography

Michael Dean obtained his Ph.D. from the Biochemistry Department at the Boston University School of Medicine. He performed his postdoctoral studies at the National Cancer Institute on the MET oncogene and cystic fibrosis gene. His laboratory's objective is to develop methods for analyzing complex diseases and to apply them to cancer and human genetic and infectious diseases. He has applied exome sequencing to detect novel and known cancer gene mutations in bladder, kidney, prostate, and adrenal tumors. Current projects include collaborations in Guatemala, Mexico, and Venezuela to study cervical cancer, HPV infection rates, retinoblastoma, and leukemia. Dean is a member of the American Society of Human Genetics, American Association of Cancer Research, Centre Etude du Polymorphisme Humaine (CEPH), the Human Genome Organization (HUGO), a Special Advisor to BGI-Shenzhen in China and an adjunct faculty member at Hood College.

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