G. Kumaramanickavel, J Clin Exp Ophthalmol 2014, 4:5
http://dx.doi.org/10.4172/2155-9570.S1.019

OMICS Group Conferences
Accelerating Scientific Discovery

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

Genomics practice in India: ophthalmic genetic diseases, DNA diagnostics, genetic counseling and research

G. Kumaramanickavel
University of Madras, India

Ophthalmic genetic diseases (OGD) are emerging as an important component of hospital outpatient departments of tertiary eye hospitals in India. The explosion of genomic knowledge and exceedingly precise, at the same time, affordable current DNA diagnostic techniques has resulted in a paradigm shift in OGD clinical practice and counseling. Patients with OGD undergo appropriate ophthalmic examination, investigations for diagnosis, which is finally accompanied with DNA diagnostics and genetic counseling. The causative genes for these disorders are nearly 600, so far identified in the human genome. India has many patients with genetic disorders who have all types of inheritances - autosomal dominant/recessive (AR) and X-linked recessive. AR inheritance is more frequent in India, compared to the Western world, due to increased practice of consanguineous marriages. Patients have obtained simple DNA genetic diagnostic services in our experience for planning their family.

Now-a-days, genetic diagnostic services have turned complex due to the availability of exome sequencing. However, there is a learning curve while interpreting the putative DNA variations and comparing it with the phenotype. We have identified the following mutations in the genes - RPGRIP1 (c.1132insA; p.Y378IfsX1), RP1 (c.5008G>G/A; p.A1670T); RPE65 (c.394G>G/A; p. A132T), ABCA4 (c.3064G>A; p.E1022K). Genetic counseling has generated good solutions with exome sequencing but has raised new social issues.

Counseling exercises lead us to map various genes in research through homozygosity mapping (HM) in these consanguineous AR families. Through HM we have identified novel genes (arrestin, RPE65, prominin like 1, FAM161A, SLC38A8) and novel mutations (Lebercilin, CERKL, BBS).

Biography

G. Kumaramanickavel graduated in medicine (MD) at the University of Madras. He was a Research Fellow at the University of Otago, New Zealand and a Visiting Associate at the National Eye Institute, NIH, USA. He is the Director of Research at Narayana Nethralaya Eye Hospital, India. For the last two and a half decades, he has been working on ocular genomics - primarily involved in gene mapping, mutational screening and association studies including genomewide in complex and Mendelian ophthalmic diseases. He has published more than 90 papers and books. He is a reviewer for many ophthalmic journals.

gkumarmvel@gmail.com