Gene Testing Reveals New Insights into Retinal Dystrophies

Synopsis: With recent access to gene testing, the genotype phenotype correlation picture is evolving. Examples of novel gene mutations causing a particular phenotype, unexpected gene mutations of a known phenotype and extreme phenotype difference within family members will be discussed. Methods of confirming a disease-causing gene defect – Polyphen, SIFT, Muttaster – will be explained.
Clinical examples of variable phenotypes in Peripherin/ RDS (PRPH2), mitochondrial mutations and their pheno- typic overlap with other non-inherited disorders, hetero- zygous ABCA4 and other diseases will be presented. Ancillary testing including electrophysiology and multi- modal imaging features aid in the process. A reasonable combined clinical and genotyping approach towards arriving at a diagnosis of a given patient in a clinical set- ting will be summarised.