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Title: Genetic characterization of syndromic and non-syndromic autosomal recessive retinal dystrophies in the Saudi population

Leen Abu Safieh
King Fahad Medical City Research Cente, Saudi Arabia

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Abstract

Background: Inherited retinal dystrophies (RD) are common cause of blindness which is characterized by loss of photoreceptor function; it contributes significantly to the global etiology of blindness especially in the industrialized world. Clinically and genetically, retinal dystrophies are known to be extremely heterogeneous. This kind of heterogeneity poses a major diagnostic challenge and makes it difficult to provide a molecular diagnostic protocol which can improve counseling and development of gene-specific treatment strategies. The Saudi population is a highly consanguineous population with many patients requesting genetic testing to identify the type of retinal dystrophy and understand the inheritance pattern in their family. Once the pathogenic mutation is identified, it is possible to identify carries and patients at risk of developing the condition in these families. Retinal dystrophies can be inherited in different patterns, however due to the nature of the Saudi population autosomal recessive is the major form of inheritance in this community. This study will aim to characterize the genetic cause underlying the syndromic and non-syndromic retinal dystrophy and use the data to understand the disease mechanism and develop genetic screening, counseling and prevention protocols.

Methodology: Patients were clinically evaluated in the ophthalmic clinic at KFMC, Princes Nora general hospital and Al Habib Hospital. Patients diagnosed with retinal dystrophy were consented and blood was collected from all available affected individuals and family members. DNA was extracted and used for whole exome sequencing following the standard protocol. Genomic variants were annotated using the ion-reporter exome analysis pipeline. To date 54 individuals representing 21 families has been recruited to this study; 12 of these were subjected to exome sequencing.

Results & Conclusion: On average 35,000 variants have been identified in each individual sequenced. Analysis filtration was performed mainly against the following criteria: homozygosity, variant location and pathogenicity likelihood. On average 10-15 variants remained in each individual. This number was reduced significantly when analyzed against the known genes involved in retinal disease. We have identified six mutations in known genes (RP1, ABCA4, RPA2, TULP1 and BBS1); further experimental validation is required to establish variant pathogenicity.

Biography

Leen Abu Safieh has completed her PhD in Human Molecular Genetics from the Institute of Ophthalmology, UCL, London, UK in 2003. She had a Master's degree in Biomedical Sciences from Westminster University, London, UK. She has over than 15 years of experience in the field of Molecular Genetics. Her research resulted in the identification of seven novel genes all involved in different retinal diseases, and all been published in high impact factor journals. Her main area of research is focused on inherited genetic diseases such as ophthalmic conditions and other complex genetic disorders. Currently she is working as a Clinical Research Consultant, at King Fahad Medical City Research Center, Saudi Arabia.