

Identification of FH causing mutations/SNPs in Malaysian population

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Abstract

Familial hypercholesterolemia (FH, ICD-10 code E78.0) is an autosomal dominant disorder that causes severe elevations in total plasma cholesterol and low density lipoprotein cholesterol (LDLC), and is associated with a high risk for premature coronary artery disease (CAD). It is caused mainly by mutations, single nucleotide polymorphisms (SNPs), large deletions or insertions in the LDLR, APOB or PCSK9 genes. Over 1,500 genetic variants have been identified in the *LDLR* gene so far, as reported by British Heart Foundation (BHF) based on studies done mainly in the Caucasian population. However, there are not many genetic variants of LDLR gene documented in Malaysian population. The current method of FH diagnosis is based on clinical criteria, which only make the diagnosis when the disease is at an advanced stage. In the present study, we are investigating the FH-causing genetic variants on LDLR in Malaysian cohort. Thus far, we have identified eight single nucleotide mutations/SNPs in clinically diagnosed FH patients by DNA sequencing. The eventual goal of the project is to devise a microarray-based diagnostic tool which will aid in the screening and early diagnosis of Malaysian FH patients, and thus providing a more effective means for disease management.

Biography

Dr. Livy Alex has completed her Ph.D in 1994 from the Department of Microbiology, Osmania University, India and was a post-doctoral fellow at the Centre for Cellular and Molecular Biology, Hyderabad, India. Thereafter she joined the biotechnology industry and worked for Sudershan Biotech Pvt., Ltd, Hyderabad(1999-2003), Reliance Life Sciences, Mumbai(2003-2007) and Actis Biologics, Mumbai, India(2007-2008). At present, she heads the Molecular Research Labs at INFOVALLEY Group of Companies (Malaysia/India). Her research interests are human genomics, recombinant proteins, molecular diagnostics, and yeast biotechnology. She has a few patents to her credit and is currently involved in developing diagnostic microarray chips for complex inherited diseases.