

## **Abstract**

The role of microRNAs (miRNA) in the control of cell growth, differentiation, apoptosis and tumorigenesis is well recognized. MicroRNAs exerts their effects by binding complementary to the 3' untranslated regions (UTR) of messengerRNAs (mRNA) and interfering with their translation. A single microRNA can target many different sites on the same mRNA or on many different mRNAs. About 3% of genes code for microRNAs and up to 30% of human protein coding genes may be regulated by miRNAs. Single Nucleotide polymorphisms (SNPs) within the miRNA targets, could interfere with the activity of miRNA's, and determine an individual's risk of developing complex diseases. The Sri Lankan Personal Genome (SLPG) was sequenced recently. The objective of this work was to identify and catalog the SNPs located in the miRNA binding sites in the SLPG.

Two MySQL relational tables were created. One is with the updated SLPG-SNP profile which contains SNPs and its location on chromosome, rsNumber if known and a serial number if it is a novel. The other table is created with the downloaded data of all predicted targets of TargetScan v5.1 for conserved miRNA families in the 3'UTR of RefSeq genes. This contain, conserved miRNA family name, target gene and targets chromosome start and end position (8nucleotides). Then a simple MySQL query executed using two above tables to select all SLPG-SNPs which are present within the targets chromosome start and end positions.

There were 85 SNPs identified in the miRNA binding sites in the SLPG. 75 were SNPs already identified in other populations. 10 were SNPs not found in other populations. An online catalogue of disease associated miRNAs in the SLPG was produced: <http://199.230.110.123:8080/lankamirnew>

SNPs located in miRNA binding sites are likely to play an important role in disease. The identification of such SNPs, which are unique to our population, in the SLPG, underscores the need to initiate research into the aetiology of all types of disease in our population because the clinical picture and treatment outcomes may differ in our population due to our unique genetic makeup.