1. Abstract

The first Sri Lankan Personal Genome (SLPG) was sequenced in 2010. SLPG was analysed to identify clinically significant single nucleotide polymorphisms (SNPs) in the SLPG.

The clinically significant SNPs associated with increased risk of diseases identified by Genome Wide Association Studies (GWAS) are found in HuGE Navigator (http://www.hugenavigator.net/HuGENavigator/downloadcentre.do). The SNP profile of the SLPG was compared with clinically significant SNPs found in the GWAS database.

There are 7095 SNPs in the HuGE Navigator database that are known to confer increased risk of various diseases in other populations. Of them 2396 (33.77%) were found in the SLPG. The number of SNPs related to genes involving disorders of various systems were as follows: cardio vascular system 619 (25.83%), nervous system 562 (23.45%), digestive system 399 (16.65%), endocrine system 240 (10%), reproductive system 168 (7%), respiratory system 78 (3.25%), musculoskeletal system 73 (3.05%), skin 66 (2.75%), renal system 48 (2%), eye 48 (2%), other 95 (3.96%).

The identification of the presence and absence of SNPs associated with increased risk of various diseases in other populations, in the SLPG, point to the possibility of similarities as well as differences in the clinical picture and treatment outcome of these conditions which underscore the need for research into these conditions in our population.