1. Abstract

The first Sri Lankan human genome of an anonymous individual from hill country origin was sequenced in year 2010. This was mapped to the build 37.2 of the reference human genome of the National Center for Biotechnology Information advances science and health (NCBI), and it was filtered for Single nucleotide polymorphisms (SNPs) using the reference human genome. Those variations were identified using build 131 of the Single Nucleotide Polymorphism Database (dbSNP). There were 2,806,370 Single Nucleotide Polymorphism Database (dbSNP) verified and 216,499 novel single nucleotide polymorphisms were found.

This study was done to compare the SNP profile of the Sri Lankan human genome with SNP profiles of five other human genomes. As the first step all the 6 SNP profiles were taken to a common comparable stage by updating their SNP profiles to the latest build (135) of the dbSNP. Novel SNPs which had been mapped to older builds of the reference human genome were remapped to the build 37. Once all SNPs were mapped to the latest build of the reference human genome, they were updated to the dbSNP build 135 using a locally created database and a simple dbSNP build upgrading mechanism. Once Sri Lankan SNP profile was updated there were total of 2,808,655 dbSNP verified and 207,934 novel SNPs. Total of 7801 build 131 novel SNPs were verified in the build 135 of the dbSNP.

Comparison of SNP profiles were done next. Here it was found that Sri Lankan genome shares more SNPs with Korean genome (SJK) (57.29%) than with other 4 genomes. (44.63% with Han Chinese in Beijing, China (CHB), 43.81% with Yoruba (YRI), 21.34% with Toscans in Italy (TSI), and 21.28% with Gujarati Indians living in Houston (GIH).) Though it shares more SNPs with Korean genome, significant difference of 1,288,327 SNPs (42.71%) was detected with the Korean genome. Also there were 663,826 SNPs that were specific to the Sri Lankan genome, and that was 22% of the total Sri Lankan SNP profile. Though the Sri Lankan SNP profile shares only 21.28% of SNPs with the Gujarati Indians (GIH) population, GIH shares 45.22% of its total SNP count with the Sri Lankan population.

These findings suggest that there are significant genetic differences between individuals from various populations.