

ABSTRACT

Background & Aim: Selection pressure has left traceable molecular selection signatures within the human genomes. Identification of selection signatures is vital for understanding the evolutionary processes shaping human adaptation and diversity in disease susceptibility. In this study, genomes of Luhya and Maasai ethnic groups were compared to identify selection signatures that may explain their genetic divergence.

Materials and Methods: The population studied composed of 90 Luhyas' and 31 Maasais' from Webuye and Kinyawa, Kenya respectively. Data were sourced from 1000 genomes project phase 3. These data were processed through similar statistical quality control protocols.

Results: Datasets were processed individually then merged; resulting in 1,480,668 high-quality SNPs. Genomic scan using F-statistics (Fst) method revealed putative genomic selection signatures. Fifty-five genomic regions (high Fst, 5.6 to 9.8). These regions overlapped with 21 genes. Three genes (*C9orf24*, *BAZ2A*, and *SYMPK*) out of the 21 are associated with the missense variants that could explain partially the genetic divergence and variation in fertility and disease susceptibility between these populations. The genes are associated with spermatogenesis (*C9orf24* gene), prostate cancer and prostatitis (*BAZ2A* gene) and vein disease (*SYMPK* gene).

Conclusion: The study suggests that the two populations have undergone diverged selection leading to variation in fertility and susceptibility to prostate cancer, prostatitis and vein disease. It highly recommended the ethnic groups should consider changing their culture to avoid challenges identified in this study.

Keywords: fertility, diseases, luhya, maasai, diversity