

Genomic characterization of HLA class I and class II genes in ethnically diverse sub-Saharan African populations: A report on novel HLA alleles

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Abstract

HLA allelic variation has been well studied and documented in many parts of the world. However, African populations have been relatively under-represented in studies of HLA variation. We have characterized HLA variation from 489 individuals belonging to 13 ethnically diverse populations from rural communities from the African countries of Botswana, Cameroon, Ethiopia, and Tanzania, known to practice traditional subsistence lifestyles using next generation sequencing (Illumina) and long-reads from Oxford Nanopore Technologies. We identified 342 distinct alleles among the 11 HLA targeted genes: HLA-A, -B, -C, -DRB1, -DRB3, -DRB4, -DRB5, -DQA1, -DQB1, -DPA1, and -DPB1, with 140 of those alleles containing novel sequences that were submitted to the IPD-IMGT/HLA database. Sixteen of the 140 alleles contained novel content within the exonic regions of the genes, while 110 alleles contained novel intronic variants. Four alleles were found to be recombinants of already described HLA alleles and 10 alleles extended the sequence content of already described alleles. All 140 alleles include complete allelic sequence from the 5' UTR to the 3' UTR that are inclusive of all exons and introns. This report characterizes the HLA allelic variation from these individuals and describes the novel allelic variation present within these specific African populations.