

## P071

## THE HLA POLYMORPHISM IN DIFFERENT NPC INCIDENT RATE REGION OF CHINA

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**Aim:** Nasopharyngeal carcinoma (NPC) has a distinct racial and geographic distribution caused by a combination of Epstein-Barr virus (EBV) infection and environmental factors. It also had been shown a strong and consistent HLA associated. The HLA genes differ greatly among populations making it as a powerful tool for the study of population genetics and disease association. We have conducted a molecular HLA typing project to describe the HLA class I profiling in the high NPC incident area of Southern China. The results also compared with reported data from Northern Chinese population previously.

**Methods:** High resolution HLA typing was informative for 629 unrelated cancer free individuals. The difference between these subjects and the former reported northern Chinese population were characterized in frequencies and in the presence of alleles and haplotypes.

**Results:** A number of 26, 52, and 21 alleles in HLA-A, B, and C loci were identified in these subjects. 21 alleles with significantly different distributions were observed in HLA-A, B, and C loci between these two groups. HLA-A\*11:01 is the most common allele with extraordinary high frequencies which indicates nature selective forces may acted on the population of NPC high incident regions.

**Conclusions:** Our study demonstrates a distinct HLA class I alleles distribution in high NPC incident region of southern Han Chinese compare with northern Han Chinese. Haplotype analyses have shown that a fewer predominant haplotype patterns in this population with higher population coverage illustrated the direction for deeply exploring by future studies.

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## P072

## HLA TYPES IN ETHNICALLY DIVERSE SUB-SAHARAN AFRICAN POPULATIONS

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**Aim:** A comprehensive characterization of the HLA allelic diversity of ethnically diverse populations of African ancestry from sub-Saharan Africa (SSA) has been attempted.

**Methods:** DNA was isolated from whole blood of 520 volunteers from 12 ethnically diverse populations in Botswana, Cameroon, Ethiopia and Tanzania. All individuals were sampled from rural communities and known to practice traditional subsistence lifestyles (hunter-gathering, agro-pastoralism or pastoralism). Current analysis is part of a larger study ( $N = 3000$ ) aimed to document genomic variation, population history, natural selection and genetic/environmental risk factors associated with diabetes, hypertension and infectious disease in African descent populations. In this preliminary study, 231 unrelated individuals were fully characterized at 11 HLA loci by next generation sequencing-based (Holotype, OMIXON) third-field resolution HLA typing.

**Results:** Among the 231 individuals, 291 HLA alleles were identified: 42 HLA-A, 52 HLA-B, 36 HLA-C, 22 HLA-DPA1, 47 HLA-DPB1, 20 HLA-DQA1, 26 HLA-DQB1, 31 HLA-DRB1, 7 HLA-DRB3, 5 HLA-DRB4 and 3 HLA-DRB5. These 291 alleles included 31 (11%) novel alleles (6 synonymous and 25 non-synonymous) and 260 (89%) previously described alleles. The populations from Eastern Africa carried the highest number of new alleles ( $n = 16$ ) compared to Southern ( $n = 10$ ) and Central ( $n = 8$ ) Africa.

**Conclusions:** This study has identified 260 known and 31 previously unknown HLA allele sequences in just 231 unrelated individuals from 12 distinct populations in SSA. These 31 previously uncharacterized alleles were quite frequent in these populations. These data provide critical information potentially enriching our understanding of the evolution of HLA polymorphisms in Africa and the role they may play in health and disease.

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