UDC 575
DOI: 10.2298/GENSR1101205R
Short communications

IDENTIFICATION OF NOVEL SINGLE NUCLEOTIDE POLYMORPHISM (SNP) IN DPB1 GENE IN ETHNIC POPULATION FROM WEST BENGAL

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Raha O., B.N. Sarkar, P. Veerraju, L. Pramanik, and V.R.Rao (2011): *Identification of novel single nucleotide polymorphism (SNP) in dpb1 gene in ethnic population from West Bengal*. - Genetika, Vol 43, No. 1, 205 -208.

HLA-DP antigens present peptides to CD4+ T cells and play an important role in autoimmune diseases and parasitic infections. We have sequenced HLA-DPB1 exon-2 from the ethnic populations in West Bengal, India and report a novel single nucleotide polymorphism (SNP) - rs111221466. The rs111221466 SNP induced silent mutation from CGA (Arg) to TGA (Stop Codon) and showed a frequency of 83.24%. In conventional sense, the frequency of novel SNP is very high. We have

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sequenced HLA-DPB1 exon-2 from a Bengali Population in West Bengal, India. HLA-DP antigens present peptides to CD4+ T cells and play an important role in autoimmune diseases and parasitic infections. Here, we report a novel single nucleotide polymorphism of HLA-DPB1 gene in the population. rs111221466 showed a frequency of 83.24, which is important to note, in view of common polymorphisms involved in disease susceptibility.

Key words: Human leukocyte antigen, Single nucleotide polymorphisms, Sequence-based typing

Human leukocyte antigen (HLA) is one of the most polymorphic genetic systems in human genome. To date, 4,447 HLA alleles have been reported according to IMGT/HLA database (ROBINSON, 2009). The DPB1 region has 138 alleles and 120 proteins. The DP sub region of the HLA class II D region contains genes encoding the alpha (142880) and beta(142858) chains of a heterodimeric, cell-surface glycoprotein that presents antigens to CD4+ (helper) T lymphocytes (VELICKOVIC, 2001). Because the HLA class II molecules are highly polymorphic, they can embrace a wide variety of antigens in their antigen-binding groove and present them to diverse T-lymphocyte antigen receptors, triggering antigen recognition. Amino acids located at key positions along the alpha-helical portions of these HLA heterodimers dictate which peptide antigens can bind (DÍAZ, 2003). Even single amino acid substitutions in these regions may alter the shape of HLA-peptide binding pocket sufficiently to change its specificity.

Blood samples were collected, with appropriate consent, from 179 individuals from ethnic Population in West Bengal, i.e., from three generation they reside in West Bengal. From each individual 5 ml of intravenous blood was collected in tubes containing EDTA. DNA was isolated using the Phenol-Chloroform procedure (SAMBROOK, 2001).

Exon 2 of DPB1 gene was amplified with the oligonucleotide primers (Forward- 5'GAGAGTGGCGCCTCCGCTCAT 3', Reverse- 5' GCCGGCCCAAAGCCCTCACTC 3') by polymerase chain reaction (PCR) in ABI GeneAmp 9700 thermalcycler (TANAKA, 1999). The PCR products were purified by Exo SAP reaction before operating the sequencing. The products were sequenced with the ABI PRISM Big Dye terminator Ready Reaction Cycle Sequencing kit on an ABI prism ^{TM} 3730. All PCR fragments were sequenced in forward primer. The sequencing results showed the sequence of the SNP differs from wild type by one nucleotide substitution (transition) in exon 2, nt 361 where C→T (codon 92 CGA →TGA); resulting in amino acid changes to stop codon, Arg→Stop (Figure 1). HLA-DPB1 showed a frequency of 83.24.The sequence of the new SNP is being submitted to GenBank in March 2010 and assigned the SNP number rs111221466.
The significance of the present finding is important with respect to common polymorphisms involved in complex disease susceptibility and the importance of validating them with other epidemiological parameters, whenever association studies are undertaken.

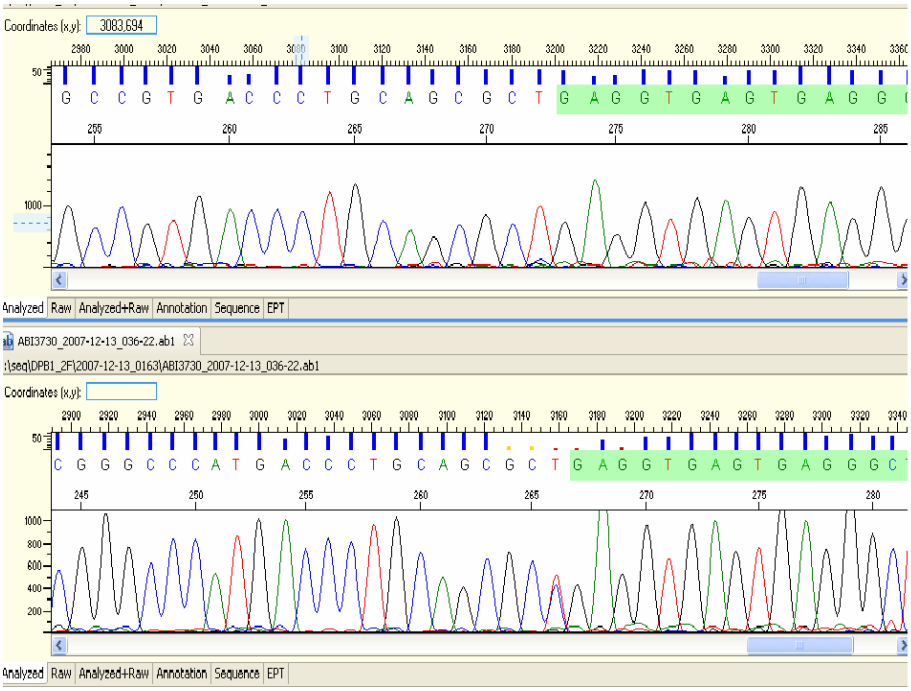

Figure 1- The figure depicts the sequence in Sequence Scanner v 1.0, showing heterozygotic condition (CT) and homozygotic mutated condition (TT).

ACKNOWLEDGMENTS

This work was sponsored by Anthropological Survey of India, Ministry of Culture, Government of India.

Received, November 18th2010
Accepted, March 14th 2011

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IDENTIFIKACIJA NOVOG POLIMORFIZMA POJEDINAČNIH NUKLEOTIDA (SNP) U DPB1 GENU ETNIČKIH POPULACIJA ZAPADNOG BENGALA

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I z v o d

HLA-DP antigeni pretstavljaju peptide prema CD4* T ćelijama I igraju važnu ulogu u autoimunim bolestima i parazitskim infekcijama. Prikaazani su rezultati eksperimenata u kojims je izvršeno sekvencioniranje HLA-DPB1 egzona – 2 u etičkim populacijama zapadnog Bengala, Indija, i utvrđen novi polimorfizam pojedinačnih nucleotide (SNP) – rs111221466.

Ovaj polimorfizam - rs111221466 SNP je indukovao tihu (silent) formu mutacije tripleta CGA (Arg) u triplet TGA (Stop Codon) sa frekvencijom od 83,24 %. U konvencijalnom smislu frekvencija polimorfzma novog pojedinačnog nukleotida (SNP) je veoma visoka. Ovo je značajno naglasiti sa aspekta normalnog polimorfizma uključenog u osetljivost prema bolestima.

Primljeno 18. XI. 2010.
Odobreno 14. III. 2011.