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HLA Immune Response Genetics

The novel HLA-DOB1 allele, HLA-DOB1*04:72, detected in a potential hematopoietic stem cell donor

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Identification of the novel HLA-DOB1*04:72 allele that differs from DOB1*04:02:01:06 by two nucleotide substitutions.

KEYWORDS HLA-DQB1, HLA-DQB1*04:72, new HLA allele, NGS

The HLA-DOB1*04:72 locus is one of the most polymorphic genes in human genome. As April 2022, a total of 2230 HLA-DOB1 alleles have been recorded in the IPD-IMGT/HLA Database.^{1,2} We describe here the identification of the new HLA-DQB1*04:72 allele, discovered during the typing of potential donors of hematopoietic stem cells recruited by National bone marrow donor registry named after Vasya Perevoshchikov, Russian Federation.

Genomic DNA was prepared from whole blood using a commercial kit, according to the manufacturer's instructions (QIAGEN, Germantown, Maryland). HLA typing for HLA-A, -B, -C, -DRB1, and -DQB1 loci were performed using commercial by Holotype kit (Omixon, Hungary). The sequencing was performed on the platform Illumina MiSeq (Illumina, San Diego, CA) and was analyzed with the HLA Twin software (version 4.1.0, Omixon, Inc., Budapest, Hungary) and IPD-IMGT/HLA Database version 3.37.0.

The novel DQB1*04:72 allele differs from DQB1*04: 02:01:06 by two nucleotide substitutions: the first in intron 1 a substitution (T > G) at position 1372 and the second in exon 3 a nonsynonmous substitution (C > T)

at position 515 (codon 140) resulting in an amino acid change (Threonine to Isoleucine) (Figure 1). The extended HLA typing of the individual with DOB1*04:72 is A*02:01:01G, 23:01:01G; C*06:02:01G, 07:01:01G; B*49: 01:01G, B*57:01:01G; DRB1*04:04:01G, 11:01:01G; DQB1 *03:01:01G.

The novel HLA-DQB1 allele was officially designated as HLA-DQB1*04:72 by the World Health Organization (WHO) Nomenclature Committee Factors of the HLA System in November 2020.² The nucleotide sequence has been submitted to GenBank with accession number MT658792 and to the IPD-IMGT/HLA Database with accession number HWS10060679.

AUTHOR CONTRIBUTIONS

Shamil Nizamov participated in the performance of the research and participated in data analysis. Raushania Gaifullina contributed to collection of the data. Elena Shagimardanova and Anastasiia Ananeva contributed to the design of the study and participated in the writing of the paper. All authors read and approved the final version of the manuscript.

Exon 3 100 AA Codon 105 110 DOB1*04:02:01:06 TG GAG CCC ACA GTG ACC ATC TCC CCA TCC AGG ACA GAG GCC CTC AAC CAC DQB1*04:72 -- --- --- --- --- --- --- --- --- --- --- --- --- ---AA Codon 115 120 125 DOB1*04:02:01:06 CAC AAC CTG CTG GTC TGC TCA GTG ACA GAT TTC TAT CCA GCC CAG ATC DOB1*04:72 --- --- --- --- --- --- --- --- --- --- --- --- --- ---AA Codon 130 135 140 DQB1*04:02:01:06 AAA GTC CGG TGG TTT CGG AAT GAC CAG GAG GAG ACA ACT GGC GTT GTG DOB1*04:72 --- --- --- --- --- --- --- --- --- -<u>-</u>--AA Codon 145 150 155 DQB1*04:02:01:06 TCC ACC CCC CTT ATT AGG AAC GGT GAC TGG ACC TTC CAG ATC CTG GTG DQB1*04:72 ____ ___ ___ ___ ___ ___ ___ ___ ___ AA Codon 160 165 170 175 DQB1*04:02:01:06 ATG CTG GAA ATG ACT CCC CAG CGT GGA GAC GTC TAC ACC TGC CAC GTG DQB1*04:72 ____ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ AA Codon 180 185 DOB1*04:02:01:06 GAG CAC CCC AGC CTC CAG AAC CCC ATC ATC GTG GAG TGG C DQB1*04:72 ____ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___

FIGURE 1 Sequence alignment of the exon 3 sequence of HLA-DQB1*04:72 compared to the most homologous allele HLA-DQB1*04:02:01:06, dashes (-) show identity to the DQB1*04:02:01:06 allele. The DNA sequence of HLA-DQB1*04:72 is identical HLA-DQB1*04:02:01:06 except in codon 140 where ACT of DQB1*04:02:01:06 is substituted by ATT in DQB1*04:72.

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CONFLICT OF INTEREST

The authors have declared no conflicting interests.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are openly available in the IPD-IMGT/HLA Database at https://www.ebi.ac.uk/ipd/imgt/hla/alleles/allele/?access ion=HLA24147.

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