

The novel HLA-DQB1 allele, *HLA-DQB1*04:72*, detected in a potential hematopoietic stem cell donor

Anastasiia Ananeva¹  | Shamil Nizamov¹  | Raushania Gaifullina^{1,2}  | Elena Shagimardanova¹ 

¹Institute of Fundamental Medicine and Biology, Kazan (Volga Region) Federal University, Kazan, Russian Federation

²Volga-Region Bone Marrow Donor Registry, Kazan, Russian Federation

Correspondence

Anastasiia Ananeva, Institute of Fundamental Medicine and Biology, Kazan (Volga Region) Federal University, Kazan, Russian Federation.
Email: nastya.ananeva@gmail.com

Funding information

Charity fund «National Bone Marrow Donors Registry named after Vasya Perevoshchikov»; Kazan Federal University Strategic Academic Leadership Program, Grant/Award Number: PRIORITY-2030

Identification of the novel *HLA-DQB1*04:72* allele that differs from *DQB1*04:02:01:06* by two nucleotide substitutions.

KEYWORDS

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

The *HLA-DQB1*04:72* locus is one of the most polymorphic genes in human genome. As April 2022, a total of 2230 *HLA-DQB1* 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 nonsynonymous 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 *DQB1*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

AA Codon		100		105		110											
DQB1*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							
DQB1*04:02:01:06	CAC	AAC	CTG	CTG	GTC	TGC	TCA	GTG	ACA	GAT	TTC	TAT	CCA	GCC	CAG	ATC	
DQB1*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	
DQB1*04:72	-	-	-	-	-	-	-	-	-	-	-	-	-T-	-	-	-	-
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								
DQB1*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*.

ACKNOWLEDGMENTS

The authors would like to thank the Charity fund «National Bone Marrow Donors Registry named after Vasya Perevoshchikov» for support of the HLA typing and for the donor recruiting. This paper has been supported by the Kazan Federal University Strategic Academic Leadership Program (PRIORITY-2030).

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/?accession=HLA24147>.

ORCID

Anastasiia Ananeva  <https://orcid.org/0000-0002-8069-7244>

Shamil Nizamov  <https://orcid.org/0000-0001-6944-1646>
 Raushania Gaifullina  <https://orcid.org/0000-0002-0922-5850>
 Elena Shagimardanova  <https://orcid.org/0000-0003-2339-261X>

REFERENCES

1. Robinson J, Barker DJ, Georgiou X, Cooper MA, Flicek P, Marsh SGE. IPD-IMGT/HLA Database. *Nucleic Acids Res.* 2020; 48:D948-D955.
2. Marsh SGE, Albert ED, Bodmer WF, et al. Nomenclature for factors of the HLA system. *Tissue Antigens.* 2010;75:291-455.

How to cite this article: Ananeva A, Nizamov S, Gaifullina R, Shagimardanova E. The novel HLA-DQB1 allele, *HLA-DQB1*04:72*, detected in a potential hematopoietic stem cell donor. *HLA.* 2022;1-2. doi:10.1111/tan.14664