

## Identification of a new *HLA-DRB1\*11* variant, *HLA-DRB1\*11:130*, by sequence-based typing in a Brazilian individual

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**Key words:** Brazilian population; *HLA-DRB1\*11:130*; new allele; sequence-based typing;

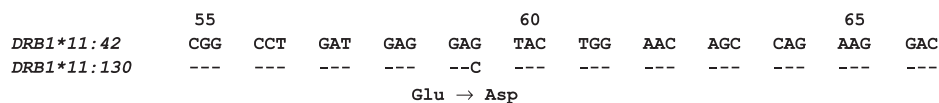
*HLA-DRB1\*11:130*, detected in a Euro-Brazilian female, presents a point mutation at codon 59.3 (GAG→GAC).

*HLA-DRB1\*11* is one of the most polymorphic *HLA-DRB1\** families, containing 186 different alleles characterized (IMGT/HLA – November 2012) (1). Here we describe a new member of this family, *HLA-DRB1\*11:130*, identified in a Euro-Brazilian female registered at the Brazilian Bone Marrow Donor Registry (REDOME).

Genomic DNA was isolated from the peripheral blood by BIOPUR kit (SR Produtos para Laboratórios Ltda, Pinhais, Brazil) and human leukocyte antigen (HLA) typing was performed using the LABType<sup>®</sup> SSOP Kits (One Lambda, Canoga Park, CA). HLA low-resolution typing for donor ID LIGH101292 was *HLA-A\*02, 66, HLA-B\*13, 18* and *HLA-DRB1\*07, 11:42*. However, the *HLA-DRB1\*11* low-resolution typing presented a false positive probe (number

28). This result prompted us to verify the possibility of mismatches between the DNA sample sequence and the probe nucleotide sequence by sequence-based typing (SBT). Subsequently, we performed SBT by AlleleSEQR Core Kit (Atria Genetics, San Francisco, CA) via ABIprism 3130 Genetic Analyzer (Applied Biosystems, Foster City, CA). Therefore, the genotype result assessment by ASSIGN software v.3.5 (Conexio Genomics, Applecross, Australia) was *HLA-DRB1\*07:01:01, 11:42* with one mismatch at exon 2. To determine which allele presents the mismatch we performed a second SBT using specific primers, AlleleSEQR *HLA-DRB1* codon 86 (Atria Genetics). This approach determined an *HLA-DRB1\*07:01:01* typing and an *HLA-DRB1\*11:42*, with a point mutation at codon 59.3, exon 2 (G→C). SBT assays were repeated and results were confirmed.

This nonsynonymous substitution results in a conservative amino acid change (GAG to GAC) with the replacement of a glutamic acid by an aspartic acid, both acidic polar amino



**Figure 1** Local sequence alignment of a partial sequence of the exon 2 of *HLADRB1\*11:42* and *HLA-DRB1\*11:130* alleles. Dashes indicate a nucleotide identical to the *DRB1\*11:42* reference sequence. Numbers above the nucleotides indicate the codon position. No nucleotide differences were found in other regions. Sequences are numbered according to the IMGT/HLA sequence database (2).

acids (Figure 1), in the  $\alpha$ -helix, adjacent to amino acids residues at the peptide contact site (3). It remains to be seen whether this conservative change would impact the peptide specificity binding.

The nucleotide sequence is available at GenBank accession number JX069944, and the World Health Organization (WHO) Nomenclature Committee officially assigned the name *DRB1\*11:130*. This follows the agreed policy that, subject to the conditions stated in the most recent Nomenclature Report, names will be assigned to new sequences as they are identified. Lists of the new allele names were published in the 2012 WHO Nomenclature Report (4).

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**Conflict of Interests**

The authors have declared no conflicting interests.

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