

Nomenclature of HLA alleles

Each HLA allele name has unique four, six or eight digit number. The length of the allele designation is dependant on the sequence of the allele and that of its nearest relative. All alleles receive at least a four digit name, six and eight digit names are only assigned when necessary.

The first two digits describe the type, which often corresponds to the serological antigen carried by an allotype, The third and fourth digits are used to list the subtypes, numbers being assigned in the order in which DNA sequences have been determined. Alleles whose numbers differ in the first four digits must differ in one or more nucleotide substitutions that change the amino acid sequence of the encoded protein. Alleles that differ only by synonymous nucleotide substitutions (also called silent or non-coding substitutions) within the coding sequence are distinguished by the use of the fifth and sixth digits. Alleles that only differ by sequence polymorphisms in the introns or in the 5' or 3' untranslated regions that flank the exons and introns are distinguished by the use of the seventh and eight digits.

In addition to the unique allele number there are additional optional suffixes that may be added to an allele to indicate its expression status. Alleles that have been shown not to be expressed, 'Null' alleles have been given the suffix 'N'. Those alleles which have been shown to be alternatively expressed which may have the suffix 'L', 'S', 'C', 'A' or 'Q'.

The suffix 'L' is used to indicate an allele which has been shown to have 'Low' cell surface expression when compared to normal levels. The 'S' suffix is used to denote an allele specifying a protein which is expressed as a soluble 'Secreted' molecule but is not present on the cell surface. A 'C' suffix to indicate an allele product which is present in the 'Cytoplasm' but not on the cell surface. An 'A' suffix to indicate 'Aberrant' expression where there is some doubt as to whether a protein is expressed. A 'Q' suffix when the expression of an allele is 'Questionable' given that the mutation seen in the allele has previously been shown to effect normal expression levels.

As of June 2008, no alleles have been named with the 'C' or 'A' suffixes.

Nomenclature	Indicates
HLA	the HLA region and prefix for an HLA gene
HLA-DRB1	a particular HLA locus i.e. DRB1
HLA-DRB1*13	a group of alleles which encode the DR13 antigen
HLA-DRB1*1301	a specific HLA allele
HLA-DRB1*1301N	a null allele
HLA-DRB1*130102	an allele which differs by a synonymous mutation
HLA-DRB1*13010102	an allele which contains a mutation outside the coding region
HLA-A*2409N	A Null allele
HLA-A*3014L	An allele encoding a protein with significantly reduced or 'Low' cell surface expression
HLA-A*24020102L	An allele encoding a protein with significantly reduced or 'Low' cell surface expression, where the mutation is found outside the coding region
HLA-B*44020102S	An allele encoding a protein which is expressed as a 'Secreted' molecule only
HLA-A*3211Q	An allele which has a mutation that has previously been shown to have a significant effect on cell surface expression, but where this has not been confirmed and its expression remains 'Questionable'