HLA CASES @CORE DIAGNOSTICS

We @CORE DIAGNOSTICS have successfully typed more than 500 cases for bone marrow transplantation. Here, through this case report we want to draw attention for new alleles as HLA genes are highly polymorphic and many substitutions occurred at positions that have not been known to be polymorphic before. A large number of HLA alleles and nucleotide variations underline the extreme diversity of the HLA system. These new alleles and their cell surface expression levels must be considered in the context of graft versus host disease as well as in the possible failure to engraft following transplantation. Specific case studies with the novel HLA alleles will be shared and potential impact to transplant outcomes evaluated.

AN INTERESTING CASE

We received the clinical sample of a patient for high resolution HLA typing who had to undergo bone marrow transplantation in the near future and had been previously diagnosed with Fanconi's anemia. Next Generation Sequence-based typing of gene HLA-A locus, showed a point mutation consisting of a nucleotide substitution of cytosine for thymine at gDNA position 800 yielding no amino acid change at codon 119 in exon 3. The closest matching reference genotype to sequence under testing was HLA-A*01:01:01:01. We released the report with typing as HLA- A*01:xx with a remark that a new allele has been observed in the patient and will be submitted to IMGT for new nomenclature. The new sequence was reported to the IMGT/HLA Database and WHO Nomenclature Committee. Subsequently, The WHO Nomenclature Committee for factors of the HLA System officially named the new sequence: **A*01:01:113.** An addendum report was released with the new officially assigned nomenclature, with prior information to the concerned clinician.

This is the regular practice that is followed @CORE DIAGNOSTICS for new allele(s) identification, submission and reporting.

REFERENCES

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