



ampli set FLT3 D835-Mt

45 tests

cat 1407

detection of D835-Mt mutation of the gene FLT3

FLT3(FMS-like receptor tyrosine kinase) is a member of the receptor tyrosine kinase class III. The receptor regulates proliferation and differentiation of haematopoietic stem cells. The FLT3 gene may present mutations that cause constitutive activation of the receptor independently of the interaction with the ligand. An Internal tandem Duplication (ITD) of the juxtamembrane (JM) domain-coding sequence of the FLT3 (FLT3/ITD) and a point mutation D835-Mt in the tyrosin-kinase domain –coding sequence of the receptor are frequently present. The ITD mutation has been identified in 20% of patients with acute myeloid leukaemia (AML) and in 3% of myelodysplastic syndrome. The point mutation D835 has been identified in 6% of AML patients. The detection of the mutations of the FLT3 gene allows a new therapeutic approach., using specific inhibitor of the tyrosine-kinase receptor. The kit allows the detection of the D835-Mt mutation, consisting of a nucleotide substitution so that the amino acid encoded isn't aspartic acid (D), but a residue of tyrosine D835Y or a residue of Valine D835V. Rarely the mutation is D835H, D835S or D835N. Whatever the mutation, it produces the lack of a restriction cleavage for the Eco RV enzyme. The detection of the mutation is carried out starting with PCR using specific oligonucleotides following by a restriction section due to Eco RV enzyme.

Principle of method: A) extraction of genomic DNA; B) amplification; C) enzymatic digestion; D) detection on agarose gel

Applicability: on extracted and purified genomic DNA from whole blood samples or tissue

ANALYSIS OF RESULTS

. The PCR product is a fragment of 114 bp. The next restriction section made by the Eco RV enzyme can be done the following results:

1	2	3
Absence of mutation	Presence of mutation	Presence of mutation
Homozygous Normal	Eterozygosis Mutant	Homozygous mutant
Patient	patient	patient
2 fragments	3 fragments	1 fragments
68 bp	114 bp	114 bp
46 bp	68 bp	
	46 bp	

REFERENCES

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