ampli set PML-RARα CE IVD 45 tests cat 1402

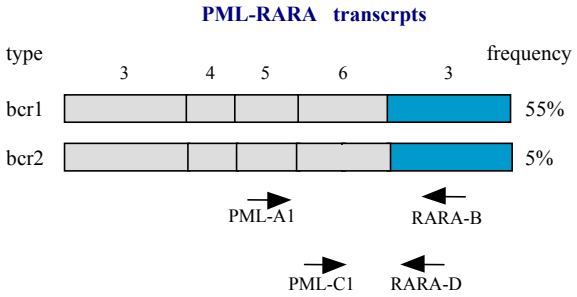
detection of translocation t(15,17)

The t(15; 17) is associated with acute promyelocitic leukemia (APL). The two genes involved in the (15; 17) traslocation are PML, coding for a putative novel transcription factor, on chromosome 15 and the retinoic acid receptor-α (RARA)gene on chromosome 17. The chromosome 17 breakpoints are localized within a 15 kb DNA fragment of the intron 2of the gene RARA . By contrast, three regions of the PML locus are involved in the translocation breakpoints: intron 6 (bcr1; 55%of cases), exon 6 (bcr2; 5 % of cases) and intron 3 (bcr3; 40% of cases). Chimeric PML-RARA and RARA-PML transcripts are formed as a consequence of the reciprocal translocation between the PML and RARA loci. The existence of different breakpoint regions in the PML locus and the presence of alternative splicing of PML are responsible for the great heterogeneity of PML-RARA junctions observed among APL patients.
The vast majority of analyzed series compared the two major PML-RARA isoforms, referred to as long (l) transcripts including PML bcr1 and bcr2 loci and short (S) transcripts (PML bcr3).The analysis with Reverse Transcription–Polymerase Chain Reaction (RT-PCR) of fusion genes is based on the design of oligonucleotide primers at opposite sides of the breakpoint fusion regions so that the PCR product contains the tumour specific fusion sequences.

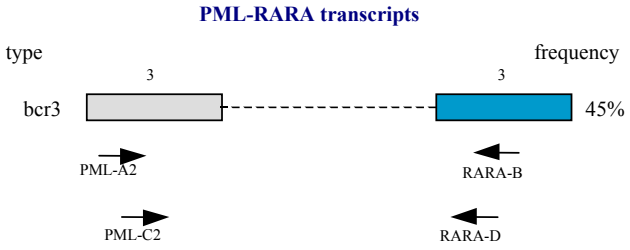
Principle of method: A) extraction of RNA; B) reverse transcription; C)amplification; D)detection on agarose gel
Applicability: on extracted and purified RNA

ANALYSIS OF RESULTS

	Size PCR products (bp)	
	I PCR	Nested PCR
bcr 1	381	214
bcr 2*	345	178



	Size PCR products (bp)	
	I PCR	Nested PCR
bcr3	376	289



The size of the PCR products could be different due to variable breakpoint positions in exon 6 of the PML gene.

REFERENCES

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