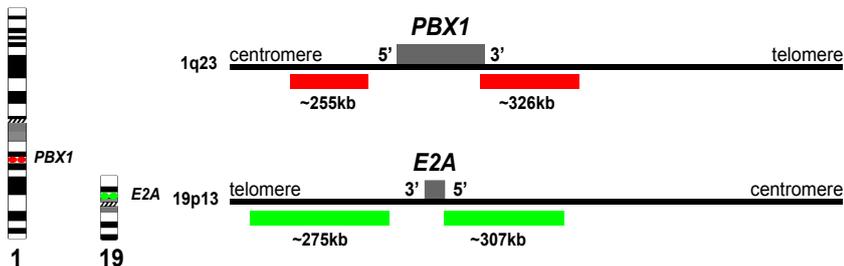


## Intended Use

The *PBX1/E2A* (also named *TCF3/PBX1*) DNA-FISH Probe is designed to detect the translocation between the *PBX1* gene located on 1q23 and the *E2A* gene located on 19p13, using fluorescence *in situ* hybridization (FISH). The translocation between the *PBX1* and *E2A* gene is designated as t(1;19)(q23;p13) and occurs in ~ 6% of pediatric and adult acute lymphoblastic leukemia (ALL) cases; as determined by conventional cytogenetics and reverse transcription-polymerase chain reaction.<sup>[1]</sup> In both pediatric and adult ALL, the translocation is correlated with a negative prognosis.<sup>[2,3]</sup> It may occur as a balanced translocation, t(1;19)(q23;p13), or as an unbalanced translocation, der(19)t(1;19)(q23;p13), where only the derivative chromosome 19 is present.<sup>[2]</sup> The unbalanced translocation, der(19), is the most common form and accounts for 75% of all *PBX1/E2A* rearrangements.<sup>[2]</sup> Both balanced and unbalanced translocations are sometimes observed in the same patient as separate clones.<sup>[2]</sup>

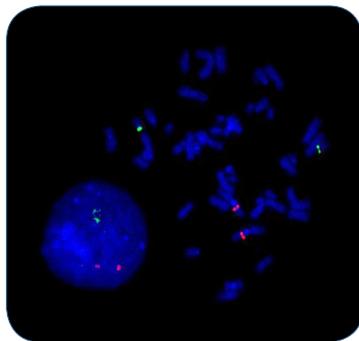


### Schematic of the *PBX1/E2A* DNA-FISH Probe:

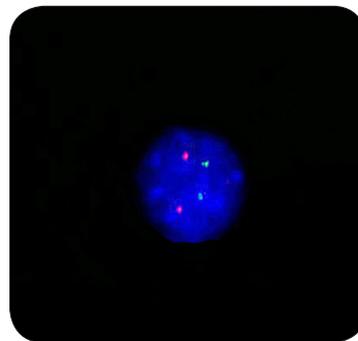
Horizontal red and green bars indicate the regions covered by the probes (approximate to scale, GRCh37/Hg19/2009). The directly labeled *PBX1* (red) & the *E2A* (green) probe flanks the respective genes.

## Signal Interpretation

In normal diploid metaphase and interphase nucleus, two red and two green signals would be observed corresponding to the two normal homologous chromosomes 1 and 19, respectively (Figures 1 and 2). Upon translocation, the most commonly observed pattern is a single red and green signal, representing the normal chromosomes 1 and 19, and two fusion signals (red/green or yellow) representing the translocated chromosomes. It is recommended to confirm variant pattern or atypical signal patterns by metaphase analysis whenever possible.



**Figure 1:** Normal diploid metaphase and interphase nucleus (from normal peripheral blood specimen) with 2 red (*PBX1*) and 2 green (*E2A*) signals.



**Figure 2:** Normal diploid interphase nucleus (from bone marrow specimen) with 2 red (*PBX1*) and 2 green (*E2A*) signals.

## References

1. Hunger, S.P., et al., *Blood*, 1991. 77(4): p. 687-93.
2. Shearer, B. M., et al. *Br J Haematol*, 2005. 129(1): p.45-52.
3. Anderson, M.K., et al., *Br J Haematol*, 2011: 155(2): p. 235-43.

## Fluorescence Microscopy Filter Requirements

Fluorophore	Excitation <sub>max</sub>	Emission <sub>max</sub>
Green	496 nm	520 nm
Red	580 nm	603 nm
DAPI	360 nm	460 nm

Instructions for use are available at [www.cancergeneticsitalia.com](http://www.cancergeneticsitalia.com)