ITALIA

Empowering Personalized Cancer Treatment

ABL1/BCR DNA-FISH Probe

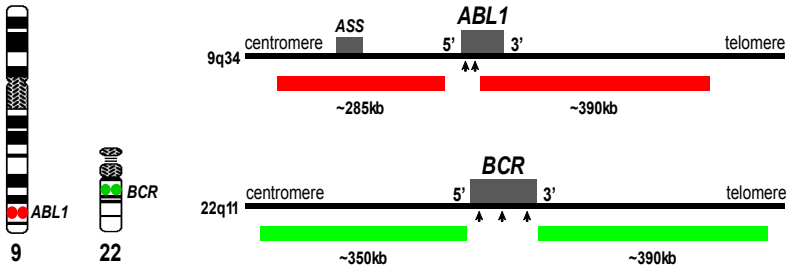
Two Color, Two Fusion Translocation Probe CE

Ref: 10-001

ABL1/BCR

Intended Use

The *ABL1/BCR* DNA-FISH Probe is designed to detect the translocation between the *ABL1* gene on chromosome 9q34 and the *BCR* gene on chromosome 22q11 by fluorescence *in situ* hybridization (FISH). This reciprocal translocation results in the Philadelphia chromosome (Ph), the der(22), and is the hallmark of chronic myeloid leukemia (CML). Approximately 90-95% of CML and up to 5% of pediatric and 20% of adult acute lymphocytic leukemia (ALL) are Ph positive.[1-3] *ABL1/BCR* FISH is used in diagnosis, prognosis, and monitoring of t(9;22) in CML and ALL patients. [4] A subset of CML (~10%) and ALL (~5%) cases exhibit large deletions adjacent to the breakpoints on chromosomes der(9) and der(22).[4-5] Such submicroscopic losses carry a poor prognosis [6] and can be detected by the Cancer Genetics Italia DNA-FISH Probe.



Schematic of the *ABL1/BCR* DNA-FISH Probe:

Horizontal red and green bars indicate the regions covered by the probes (approximate to scale, NCBI Build 36.1/Hg18/2006). The directly labeled *ABL1* (red) and *BCR* (green) probes flank the common translocation breakpoints (arrows). Breakpoints in *ABL1* can occur within a >300 kb region, often between exons 1b and 1a (arrows), and sometimes proximal to exon 1b or distal to 1a. In *BCR*, the majority of breakpoints cluster within a 5.8 kb region between exon 12-16 (*m-BCR*, middle arrow). In a subset of CML and ALL cases, the breakpoints cluster between exon 1 and 2 (*m-BCR*, left arrow). A third breakpoint cluster (*u-BCR*, right arrow) occurs distal to exon 19.

Signal Interpretation

In normal diploid metaphase chromosomes and interphase nuclei, the probe generates two red and two green signals corresponding to the two normal homologous chromosomes 9 and 22, respectively (Figure 1). In cells with translocation between *ABL1* and *BCR*, the most commonly observed pattern is one red and one green signal, representing the normal chromosomes 9 and 22, and two fusion signals (red/green or yellow) representing the two translocated chromosomes (Figure 2). Deletions adjacent to breakpoints on chromosomes der(9) and der(22) may result in variant signal patterns, most commonly a loss or reduction in brightness of one fusion signal. Variant, masked, or 3-way translocations have been reported; hybridization to tumor metaphase chromosomes is recommended to characterize the abnormal variant signal patterns.

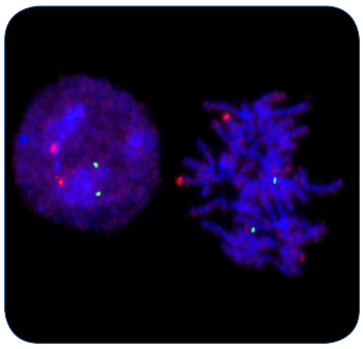


Figure 1: Normal diploid metaphase and interphase nucleus (from normal peripheral blood specimen) with 2 red (*ABL1*) and 2 green (*BCR*) signals.

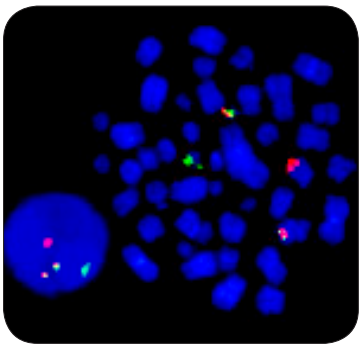


Figure 2: Interphase nuclei with 1 red (*ABL1*), 1 green (*BCR*), and 2 fusion (red/green or yellow) signals.

References

1. Huret, J. L. t(9;22)(q34;q11) in CML, Dec. 1997. www.AtlasGeneticsOncology.org.
2. Huret, J. L. t(9;22)(q34;q11) in ALL, Sep 1997. www.AtlasGeneticsOncology.org.
3. Nashed, A. L., et al. J Mol Diagn, 2003. 5:63-72.
4. Landstrom, A.P., Tefferi, A. Leuk Lymphoma, 2006. 47(3): 397-402.
5. Gorusu, M., et al. Cancer Genet Cytogenet, 2007. 173:97-106.
6. Huntly, B.J. et al. Blood, 2003. 120(4): 1160-8.

Fluorescence Microscopy Filter Requirements

Fluorophore	Excitation $_{max}$	Emission $_{max}$
Green	496 nm	520 nm
Red	580 nm	603 nm
DAPI	360 nm	460 nm

Instructions for use are available at www.cancergeneticsitalia.com

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