# Zyto Light ® SPEC PAX5 Dual Color Break Apart Probe



# **Background**

The ZytoLight® SPEC PAX5 Dual Color Break Apart Probe is designed to detect rearrangements involving the chromosomal region 9p13.2 harboring the PAX5 (paired box 5, a.k.a. BSAP) gene.

The transcription factor PAX5 activates crucial genes for B-cell lineage differentiation and represses genes that play an important role in other hematopoietic lineages. PAX5 is also implicated in human B-cell malignancies, as it is deregulated by chromosomal translocations in a subset of acute lymphoblastic leukemias (ALL). B-progenitor ALL (B-ALL), a common pediatric malignancy, is characterized by the participation of PAX5 in specific chromosomal rearrangements that generate novel fusion proteins. All PAX5 fusion proteins contain the PAX5 DNA-binding domain and thus are predicted to retain the ability to bind to PAX5 transcriptional targets, but no longer provide normal transcriptional regulatory functions. The fusion proteins contribute to B-ALL formation by competitively inhibiting the transcriptional activation of wildtype PAX5.

PAX5 rearranged ALL patients were shown to respond well to treatment with prednisone. Hence, the identification of PAX5 rearrangements by FISH may be of therapeutic significance in ALL.

Restairces

Busslinger M, et al. (1996) Proc Natl Acad Sci U S A 93: 6129-34.

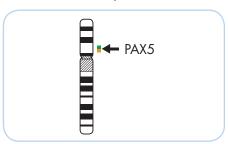
Cobaleda C, et al. (2007) Nat Immunol 8: 463-70.

Coyaud E, et al. (2010) Blood 115: 3089-97. Mullighan CG, et al. (2007) Nature 446: 758-64. Nebral K, et al. (2009) Leukemia 23: 134-43. Offit K, et al. (1992) Blood 80: 2594-9.

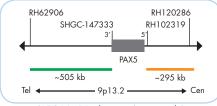
## **Probe Description**

The ZytoLight® SPEC PAX5 Dual Color Break Apart Probe is composed of:

- · ZyGreen (excitation 503 nm/emission 528 nm) labeled polynucleotides (~10 ng/µl), which target sequences mapping in 9p13.2\*\* (chr9:36,331,787-36,837,502) distal to the PAX5 breakpoint region.
- · ZyOrange (excitation 547 nm/emission 572 nm) labeled polynucleotides (~4.5 ng/µl), which target sequences mapping in 9p13.2\*\* (chr9:37,043,219-37,336,413) proximal to the PAX5 breakpoint region.
- · Formamide based hybridization buffer



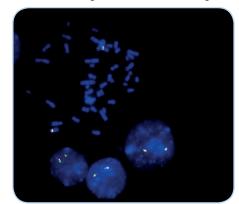
Ideogram of chromosome 9 indicating the hybridization locations.



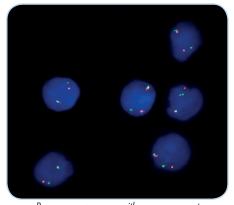
SPEC PAX5 Probe map (not to scale).

### Results

In an interphase nucleus of a normal cell lacking a translocation involving the 9p13.2 band, two orange/green fusion signals are expected representing two normal (non-rearranged) 9p13.2 loci. A signal pattern consisting of one orange/green fusion signal, one orange signal, and a separate green signal indicates one normal 9p13.2 locus and one 9p13.2 locus affected by a translocation or inversion. Isolated orange signals are the result of deletions distal to the PAX5 breakpoint region or are due to unbalanced translocations affecting this chromosomal region.



SPEC PAX5 Dual Color Break Apart Probe hybridized to normal interphase cells as indicated by two orange/green fusion signals per nucleus and to metaphase chromosomes of a normal cell.



Bone marrow smear with rearrangement affecting the PAX5 gene as indicated by one non-rearranged orange/green fusion signal, one orange and one separate green signal.

Prod. No.	Product	Label	Tests* (Volume)
Z-2300-50	Zyto <i>Light</i> SPEC PAX5 Dual Color Break Apart Probe C € [IVD]	•/•	5 (50 µl)
Related Products			
Z-2099-20	Zyto Light FISH-Cytology Implementation Kit C € IVD Incl. Cytology Pepsin Solution, 4 ml; 20x Wash Buffer TBS, 50 ml; 10x MgCl <sub>2</sub> , 50 ml; 10x PBS, 50 ml; Cytology Stringency Wash Buffer SSC, 500 ml; DAPI/DuraTect-Solution, 0.8 ml		20

<sup>\*</sup> Using 10 µl probe solution per test. 🚾 labeled products are only available in certain countries. All other countries research use only! Please contact your local dealer for more information. \*\*According to Human Genome Assembly GRCh37/hg19

Molecular diagnostics simplified FE169-1-23