ZytoLight® SPEC RUNX1/RUNX1T1 Dual Color Dual Fusion Probe

Intended Purpose

The Zyto*Light* SPEC RUNX1/RUNX1T1 Dual Color Dual Fusion Probe is designed to detect the specific translocation involving the chromosomal region 21q22.12 harboring the RUNX1 (a.k.a. AML1) gene and the chromosomal region 8q21.3 harboring the RUNX1T1 (a.k.a. ETO, CBF2T1) gene.

The balanced chromosomal translocation t(8;21) is found in about 90% of acute myeloid leukemia (AML) patients. AML is a heterogeneous clonal disorder of hematopoietic progenitor cells and one of the most common malignant myeloid disorders in adults.

The runt related transcription factor 1 gene (RUNX1) and RUNX1 translocation partner 1 (RUNX1T1) gene are both involved in the transcriptional regulation of genes during normal hematopoiesis. The non-random translocation t(8;21) (q21.3;q22.1) is strongly associated with the French-American-British (FAB) phenotype M2 (AML-M2) and produces a chimeric gene consisting of the 5'-region of the RUNX1 gene fused to the 3'-region of the RUNX1T1 gene. The chimeric protein is thought to be associated with the nuclear corepressor/histone deacetylase complex to block hematopoietic differentiation.

Fluorescence *in situ* hybridization (FISH) can provide important infor*Mation* for the management of patients with hematologic disorders.

References

Dayyani F, *et al.* (2008) Blood 111: 4338-47.
Estey E & Döhner H (2006) Lancet 368: 1894-907.
Gmidène A, *et al.* (2011) Med Oncol 28 Suppl 1: 509-12.
Licht D (2001) Oncogene 20: 5560-79.
Vangala RK, *et al.* (2003) Blood 101: 270-7.

Probe Description

The Zyto*Light* SPEC RUNX1/RUNX1T1 Dual Color Dual Fusion Probe is composed of:

- ZyGreen (excitation 503 nm/emission 528 nm) labeled polynucleotides (~12,0 ng/µl), which target sequences mapping in 21q22.11-q22.12* (chr21:35,530,283-36,855,548) harboring the RUNX1 gene region (see Fig. 1).
- ZyOrange (excitation 547 nm/emission 572 nm) labeled polynucleotides (~6 ng/µl), which target sequences mapping in 8q21.3-q22.1* (chr8:92,632,490-93,746,043) harboring the RUNX1T1 region (see Fig. 1).
- Formamide based hybridization buffer

* according to Human Genome Assembly GRCh37/hg19

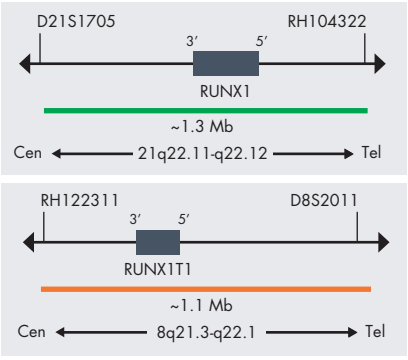
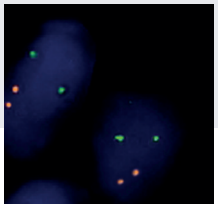


Fig. 1: Top: SPEC RUNX1 Probe map; Bottom: SPEC RUNX1T1 Probe map (not to scale)

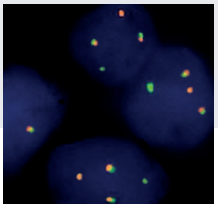
Ideograms of chromosome 21 and 8 indicating the hybridization locations

Results

Normal situation: In Interphases of normal cells or cells without a translocation involving the respective gene regions, two separate green and orange signals appear.
Aberrant situation: A gene fusion is indicated by one separate orange signal, one separate green signal, and two orange/green fusion signals.



Hybridized to normal interphase cells as indicated by two orange and two green signals in each nucleus.



Bone marrow biopsy section with translocation affecting the RUNX1/RUNX1T1 locus as indicated by one separate orange signal, one separate green signal, and two orange/green fusion signals.

Labels	Chromosome	Reg. Status	Tests	Order No.
● | ●	8,21	CE/IVD	5 (50 µl)	**Z-2112-50**
			20 (200 µl)	**Z-2112-200**