

# ZytoLight® SPEC GATA2/MECOM Dual Color Dual Fusion Probe

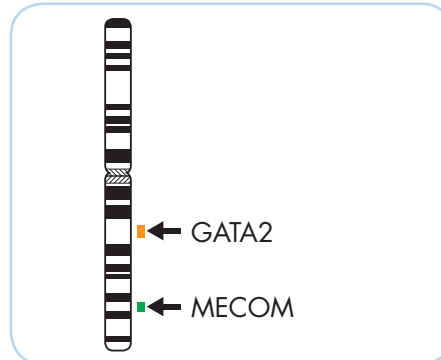


## Background

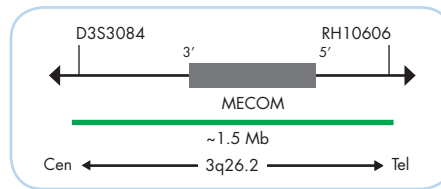
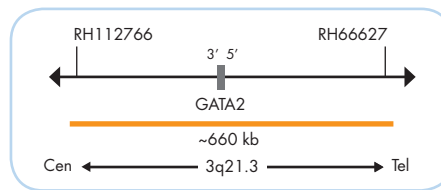
The ZytoLight® SPEC GATA2/MECOM Dual Color Dual Fusion Probe is designed to detect the inversion  $inv(3)(q21q26.2)$  and the translocation  $t(3;3)(q21;q26.2)$  both affecting the GATA2 (a.k.a. NFE1B) gene in the chromosomal region 3q21.3 and the MECOM (MDS1 and EVI1 complex locus, a.k.a. MDS1, EVI1) gene in 3q26.2. MECOM and GATA2 are transcription factors that play an essential role in the proliferation of hematopoietic stem cells.  $Inv(3)/t(3;3)$ , and less commonly  $ins(3;3)(q26.2;q21q26.2)$ , occur in 1-2.5% of acute myeloid leukemia (AML) and are also observed in myelodysplastic syndromes and in the blastic phase of chronic myeloid leukemia. A variety of other MECOM translocations involving other fusion partner genes have also been reported in various types of myeloid malignancies. 3q26.2 rearrangements are associated with minimal to no response to chemotherapy and poor clinical outcome. The  $inv(3)$  and  $t(3;3)$  result in overexpression of the MECOM gene due to its juxtaposition to enhancer sequences of the GATA2 gene and simultaneously confer GATA2 haploinsufficiency, leading to leukemogenesis. In the revised 2016 WHO classification of myeloid neoplasms and acute leukemia, "AML with  $inv(3)(q21.3q26.2)$  or  $t(3;3)(q21.3;q26.2)$ ; GATA2, MECOM" is classified as its own entity, emphasizing the unique clinicopathologic features and poorer prognosis of this subgroup of AML patients. Chromosome 3q26.2 rearrangements may be cryptic on standard karyotype analysis. Hence, FISH may be a helpful tool to confirm the diagnosis of this distinct AML subgroup.

## Probe Description

The SPEC GATA2/MECOM Dual Color Dual Fusion Probe is a mixture of an orange fluorochrome direct labeled probe spanning the GATA2 gene region at 3q21.3 and a green fluorochrome direct labeled probe spanning the MECOM gene region at 3q26.2.



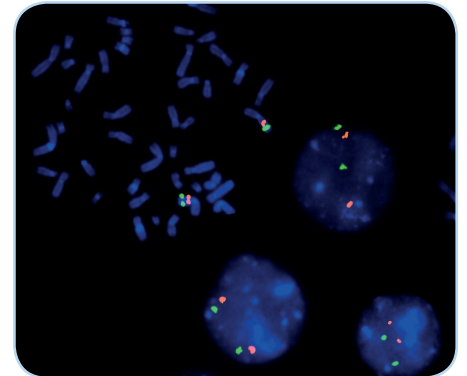
Ideogram of chromosome 3 indicating the hybridization locations.



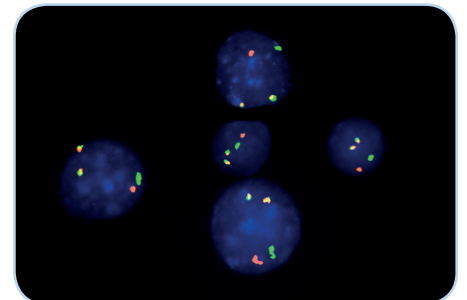
**References**  
 Arber DA, et al. (2016) Blood 127: 2391-405.  
 Bobadilla D, et al. (2007) Br J Haematol 136: 806-13.  
 De Braekeleer E, et al. (2011) Anticancer Res 31: 3441-8.  
 Pintado T, et al. (1985) Cancer 55: 535-41.  
 Tang Z, et al. (2019) Cancer Genet 233-234: 21-31.  
 Yamazaki H, et al. (2014) Cancer Cell 25: 415-27.

## Results

In a normal interphase nucleus, two green and two orange signals are expected. An aberration involving the chromosomal regions of GATA2 and MECOM generates a fusion signal on each of the chromosomes involved in case of a  $t(3;3)$  or two fusion signals on the involved chromosome in case of an  $inv(3)$ . The chromosomal regions that are not translocated are indicated by the single green and orange signal, respectively. Other relevant signal patterns may also be observed as a result of  $ins(3;3)$  or 3q26.2 rearrangements without the involvement of the GATA2 locus.



SPEC GATA2/MECOM Dual Color Dual Fusion Probe hybridized to normal interphase cells as indicated by two orange and two green signals and to metaphase chromosomes of a normal cell.



Bone marrow smear with rearrangement affecting the GATA2/MECOM loci as indicated by one separate orange signal, one separate green signal, and two orange/green fusion signals.

Prod. No.	Product	Label	Tests* (Volume)
Z-2287-50	ZytoLight SPEC GATA2/MECOM Dual Color Dual Fusion Probe	●/●	5 (50 µl)
<b>Related Products</b>			
Z-2028-5	ZytoLight FISH-Tissue Implementation Kit Incl. Heat Pretreatment Solution Citric, 150 ml; Pepsin Solution, 1 ml; Wash Buffer SSC, 210 ml; 25x Wash Buffer A, 50 ml; DAPI/DuraTest-Solution, 0.2 ml		5
Z-2099-20	ZytoLight FISH-Cytology Implementation Kit 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; Cytology Wash Buffer SSC, 500 ml; DAPI/DuraTest-Solution, 0.8 ml		20

\* Using 10 µl probe solution per test. only available in certain countries. All other countries research use only! Please contact your local dealer for more information. Please check IVD / registration status in your country

ZytoLight® FISH probes are direct labeled using the unique ZytoLight® Direct Label System II providing improved signal intensity. Advanced specificity of the single copy SPEC probes is obtained by the unique ZytoVision® Repeat Subtraction Technique.