XL t(3;3) GATA2/ MECOM DF

Translocation/Dual Fusion Probe

Order No.:
D-5124-100-OG

Description

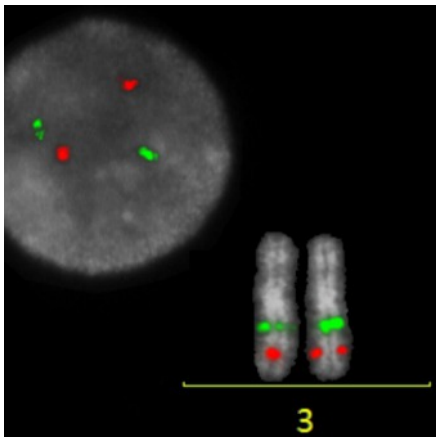
XL t(3;3) GATA2/MECOM DF is designed as a dual fusion probe. The orange labeled probe spans the breakpoint at 3q26 (MECOM), the green labeled probe spans the breakpoint at 3q21 (GATA2 and RPN1).

Clinical Details

The chromosomal aberrations inv(3)(q21q26.2) and t(3;3)(q21;q26.2) characterize a distinct entity within patients with acute myeloid leukemia (AML). Their incidence in AML is about 1%-2.5% and patients have an unfavorable prognosis and low response to chemotherapy. Inv(3)/t(3;3) juxtapose the GATA2 enhancer with the MECOM locus which results in overexpression of EVI1. The EVI1 gene is located in 3q26 and is involved in hematopoietic stem cell maintenance. EVI1 is together with MDS located in the ´MDS1 and EVI1 complex locus´ (MECOM) and is the major player in this subtype of AML. Furthermore, structural rearrangements caused by inv(3)/t(3;3) result in reduced GATA2 expression which may contribute to the oncogenic potential of this aberration. GATA2 is located in chromosomal region 3q21 and is involved in development and proliferation of hematopoietic stem cells. Several other recurrent 3q26 rearrangements as t(3;21)(q26;q22), t(3;12)(q26;p13) and t(2;3)(p15-23;q26) are known and many more rearrangements may occur.

Literature:

- Gröschel et al (2014) Cell 157:369-381
- De Braekeleer (2015) Fut Oncol 11:1675-1686
- Arber et al (2016) Blood 19:2391-2405



XL t(3;3) GATA2/MECOM DF hybridized to lymphocytes. One normal interphase and two chromosomes 3 are shown. The expected normal signal pattern of XL t(3;3) GATA2/MECOM DF is two orange and two green signals, representing the two normal MECOM and GATA2 loci. Both, inv(3)(q21q26.2) and t(3;3)(q21;q26.2), result in a split of the green (GATA2) and the orange signal (MECOM), resulting in two green-orange colocalization/fusion signals. The remaining normal copies of MECOM and GATA2 are contributing one orange and green signal each. Reciprocal translocations with unknown partner chromosomes result in a signal constellation of two green and three orange signals. A deletion of the long arm of chromosome 3 is indicated by one green and one orange signal. Other signal patterns may occur.

Clinical Applications:

- AML
- MDS

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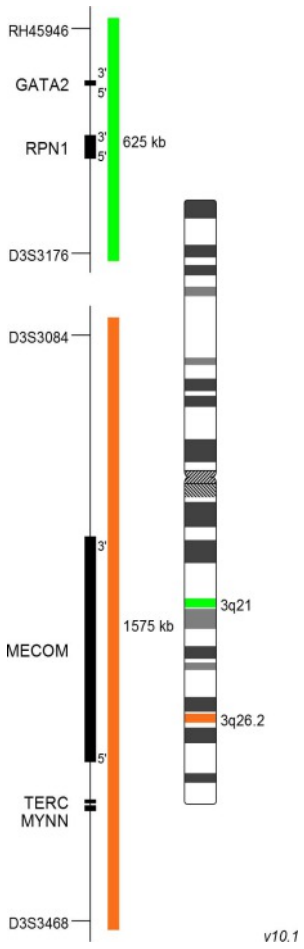
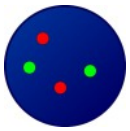
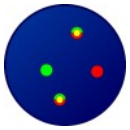
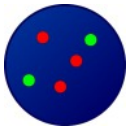
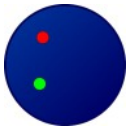


Normal cell: The expected signal pattern is two green and two orange signals, 2G2O. The probe design might result in a higher number of green-orange colocalization signals in normal cells than observed with translocation probes located on different chromosomes.	
Aberrant cell: Both, inv(3)(q21q26.2) and t(3;3)(q21;q26.2), result in the signal pattern one green, one orange and two colocalization/fusion signal, 1G1O2GO.	
Aberrant cell: A reciprocal translocation with an unknown partner chromosome results in the signal pattern two green and three orange signals, 2G3O.	
Aberrant cell: A deletion of the labeled region in the long arm of chromosome 3 results in the signal pattern one green and one orange signal, 1G1O.	

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