

XL
CBFB/MYH11
plus
Translocation/Dual Fusion
Probe

Order No.:
D-5126-100-OG

Description

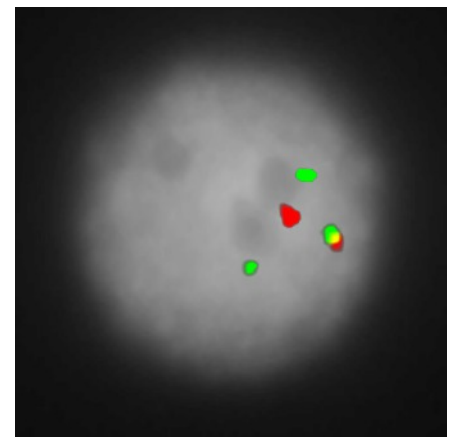
XL CBFB/MYH11 plus is designed as a dual fusion probe. The orange labeled probe spans the breakpoint at 16q22 (CBFB), and the green labeled probe spans the breakpoint at 16p13 (MYH11).

Clinical Details

Acute myeloid leukemia with $inv(16)(p13.1;q22)$ and $t(16;16)(p13.1;q22)$ is listed in the World Health Organization classification of tumors of the haematopoietic and lymphoid tissues. These recurrent rearrangements are present in about 10% of young AML patients. In cases with $inv(16)/t(16;16)$, the core binding factor b (CBFB) gene on 16q22 is fused with the smooth muscle myosin heavy chain gene (MYH11) on 16p13.1. Patients carrying $inv(16)/t(16;16)$ usually have a good prognosis. Cryptic insertions with no indication in cytogenetic analyses have been published. In these cases, a partial insertion of MYH11 into CBFB, or a partial insertion of CBFB into the MYH11 gene was observed. FISH probes with a break-apart design might overlook this cryptic rearrangement because no separation of flanking regions of CBFB occurs whereas translocation/dual fusion FISH probes are indicating this kind of cryptic rearrangement. FISH is a complementary method for the detection of $inv(16)/t(16;16)$ increasing the sensitivity in combination with conventional cytogenetics. Furthermore, FISH is a valuable tool for cases without assessable metaphases.

Literature:

- Fröhling et al (2005) Haematologica 90:194-199
- Van Obbergh et al (2014) Cancer Genetics 207:231-232
- Zhang et al (2017) Adv in Mod Onc Res 3:12-14

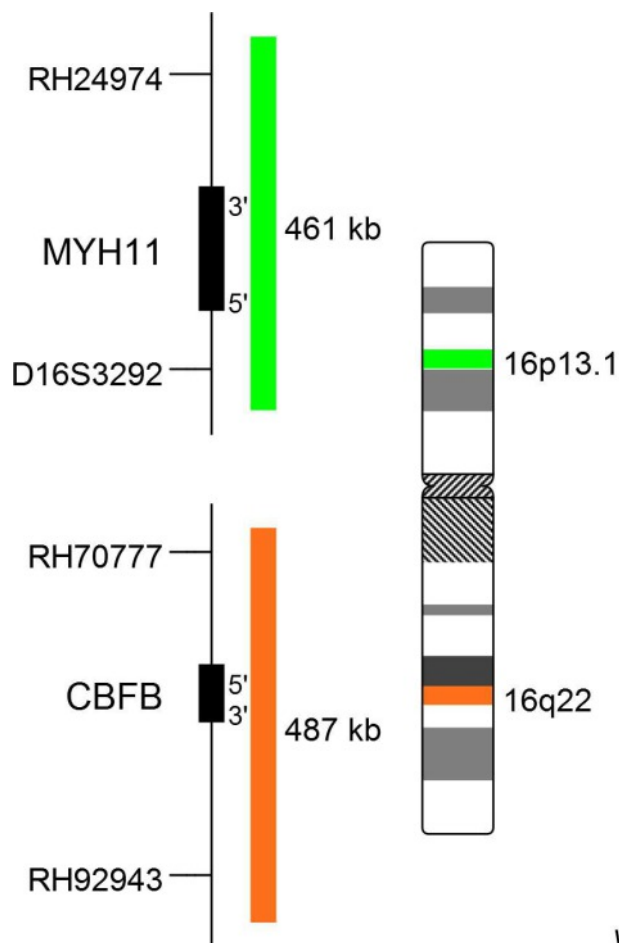


XL CBFB/MYH11 plus hybridized to bone marrow cells. One aberrant interphase is shown. The expected normal signal pattern of XL CBFB/MYH11 is two green and two orange signals, representing the two normal CBFB and MYH11 loci. The image above shows the relatively rare case of a partial insertion of MYH11 into CBFB. The aberrant signal pattern of this cryptic insertion is two green, one orange and one colocalization/fusion signal. The most common aberrations are the pericentric inversion $inv(16)$ and the reciprocal translocation $t(16;16)$ with breakpoints in CBFB and MYH11. This causes a split of the orange and green signal, resulting in two green-orange colocalization/fusion signals. The remaining normal copies of CBFB and MYH11 are contributing one green and orange signal each.

Clinical Applications:

- AML

FACTSHEET



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Normal cell: The expected signal pattern is two green and two orange signals, 2G2O.	
Aberrant cell: Both, inv(16) and t(16;16), result in the signal pattern one green, one orange and two colocalization/fusion signal, 1G1O2GO.	
Aberrant cell: An insertion of MYH11 (16p13.1) into CFBF (16q22) results in the signal pattern two green, one orange and one colocalization/fusion signal, 2G1O1GO.	
Aberrant cell: An insertion of CFBF (16q22) into MYH11 (16p13.1) results in the signal pattern one green, two orange and one colocalization/fusion signal, 1G2O1GO.	

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