XL t(11;19) KMT2A/ MLLT1 DF

Translocation/
Dual Fusion Probe

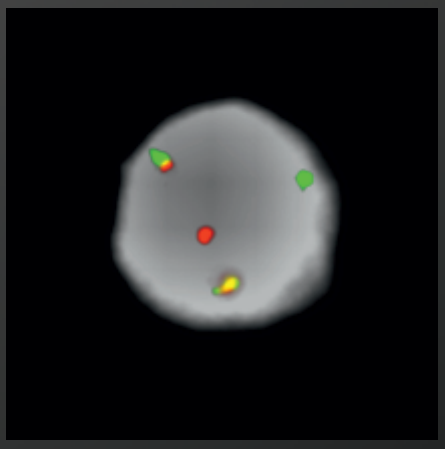
Order No.:
D-5136-100-OG

Description

XL t(11;19) KMT2A/MLLT1 DF is designed as a dual fusion probe. The orange labeled probe is located in band 19p13.3 (MLLT1), the green labeled probe spans the break-point at 11q23.3 (KMT2A).

Clinical Details

Chromosomal rearrangements of the KMT2A (lysine methyltransferase 2A) gene, formerly MLL (mixed lineage leukemia), are associated with various hematological disorders. Most patients suffer from acute myeloid leukemia (AML) or acute lymphoblastic leukemia (ALL), while only a minority develops mixed lineage leukemia (MLL). Several chromosomal aberrations involving the KMT2A gene have been identified. However, the majority of leukemias result from translocations leading to KMT2A fusions. More than 90 KMT2A translocation partner genes fused to the 5´-KMT2A portion have been identified. The most common translocation partners in KMT2A associated leukemia are AFF1, MLLT3, MLLT1, MLLT10, ELL and AFDN, described here in the order of their frequency. MLLT1 (MLLT1 super elongation complex subunit), originally designated as ENL, is one of the most common KMT2A fusion partners. KMT2A-MLLT1 fusions result from translocations of the type t(11;19)(q23;p13.3). Most patients carrying a KMT2A-MLLT1 fusion have breakpoints in intron 11 of the KMT2A gene. KMT2A-MLLT1 fusions are prevalent in both AML and ALL. Multiple KMT2A translocation partner genes, including MLLT1, are organized within the DOT1L transcriptional complex. AFF1 serves as DOT1L complex docking platform for MLLT1 and MLLT3, while MLLT10 directly mediates this interaction. More precisely, the MLLT1 protein has been shown to interact with AFF1 via its C-terminus. Due to the close interaction of these KMT2A fusion partners with DOT1L, DOT1L inhibitors are considered as promising candidates for treatment of such leukemia cases.



XL t(11;19) KMT2A/MLLT1 DF hybridized to bone marrow cells, one aberrant cell is shown. Translocations are typically observed as one orange and one green signal clearly separated and two orange-green colocalization/fusion signals.

Clinical Applications

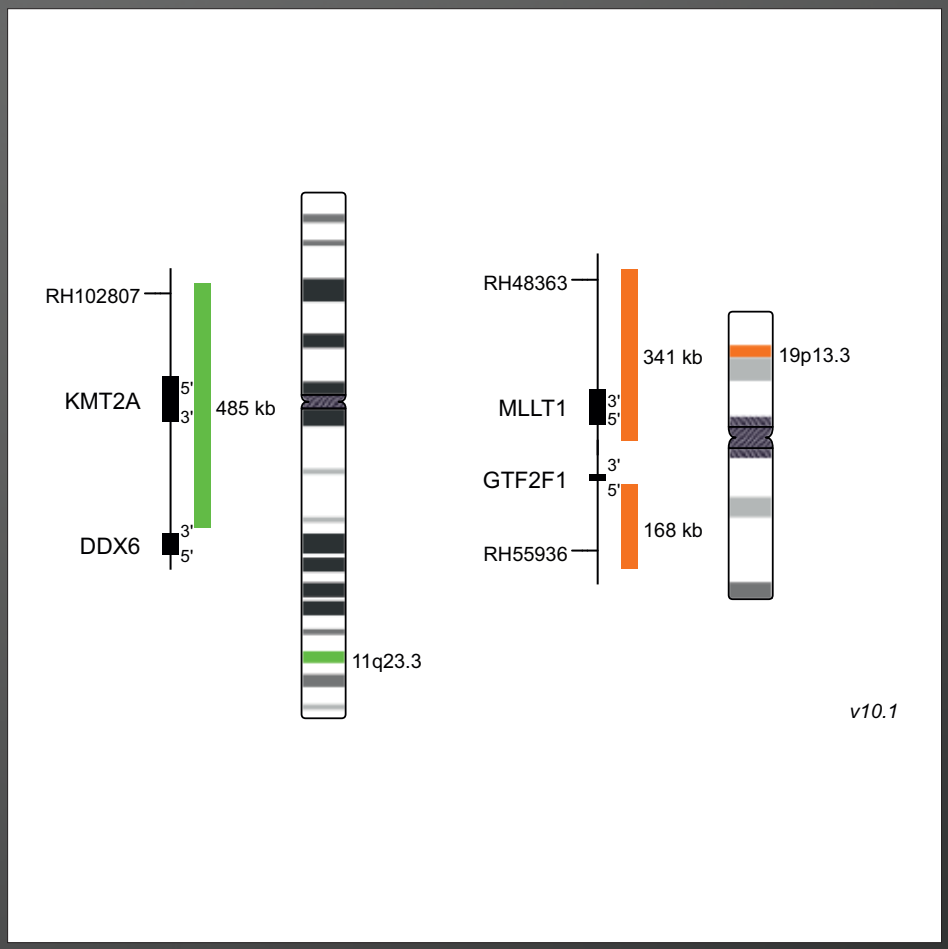
- Acute Lymphoblastic Leukemia (ALL)
- Acute Myelogenous Leukemia (AML)

Literature

- Meyer et al (2005) Proc Natl Acad Sci USA 102:449-454
- De Braekeleer et al (2011) Mol Oncol 5: 555-563
- Winters and Bernt (2017) Front Pediatr 5 (4):doi:10.3389/fped.2017.00004

FACTSHEET

MetaSystems
Probes



MetaSystems Probes

Normal Cell: Two green (2G) and two orange (2O) signals.	
Aberrant Cell (typical results): One green (1G), one orange (1O), and two green-orange colocalization/fusion signals (2GO) resulting from a reciprocal translocation between the respective loci.	

Note: In case of t(11;19)(q23.3;p13.1) with fusion of KMT2A and ELL, one orange and one green signal may appear fused on metaphase spreads due to the close proximity of ELL and MLLT1.

EUROPE & RUSSIA

Germany, Altlussheim
info@metasystems-probes.com

Italy, Milano
info@metasystems-italy.com

Russia, Moscow
info@metasystems.su

AMERICA

USA, Newton
info@metasystems.org

Argentina, Buenos Aires
info@metasystems-latam.com

ASIA & INDIA

China, Hong Kong
info@metasystems-asia.com

China, Taizhou
info@metasystems-china.com

India, Bangalore
info@metasystems-india.com

Document No. PFS-MP-D5136-EN-2020-04-01-W

FACTSHEET

MetaSystems Probes

info@metasystems-probes.com
www.metasystems-probes.com