XL t(11;19) KMT2A/ELL DF

Translocation/
Dual Fusion Probe

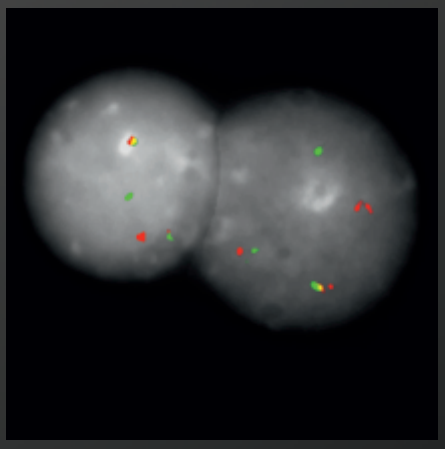
Order No.:
D-5135-100-OG

Description

XL t(11;19) KMT2A/ELL DF is designed as a dual fusion probe. The orange labeled probe is located in band 19p13.1 (ELL), the green labeled probe spans the breakpoint 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. Fusions between KMT2A and ELL (elongation factor for RNA polymerase II), caused by translocations of the type t(11;19)(q23;p13.1), belong to the most common KMT2A fusion genes in AML. Approximately 11% of AML patients carrying KMT2A rearrangements are characterized by the KMT2A-ELL fusion gene. The breakpoints within the KMT2A gene resulting in KMT2A-ELL fusions are found in intron 9 in the case of patients younger than one year and in intron 11 in the case of patients older than one year. This breakpoint distribution is unique among all KMT2A fusions. The outcome of patients with breakpoints in KMT2A intron 11 is worse compared to patients with upstream breakpoints. ELL is a component of the super elongation complex (SEC). Chimeric KMT2A-ELL fusion proteins have the ability to recruit SEC resulting in aberrant gene expression.



XL t(11;19) KMT2A/ELL DF hybridized to bone marrow cells, two aberrant cells are 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

- De Braekeleer et al (2011) Mol Oncol 5: 555-563
- Meyer et al (2017) Leukemia 32: 273–284
- Chan and Chen (2019) Front Cell Dev:doi:10.3389/fcell.2019.00081

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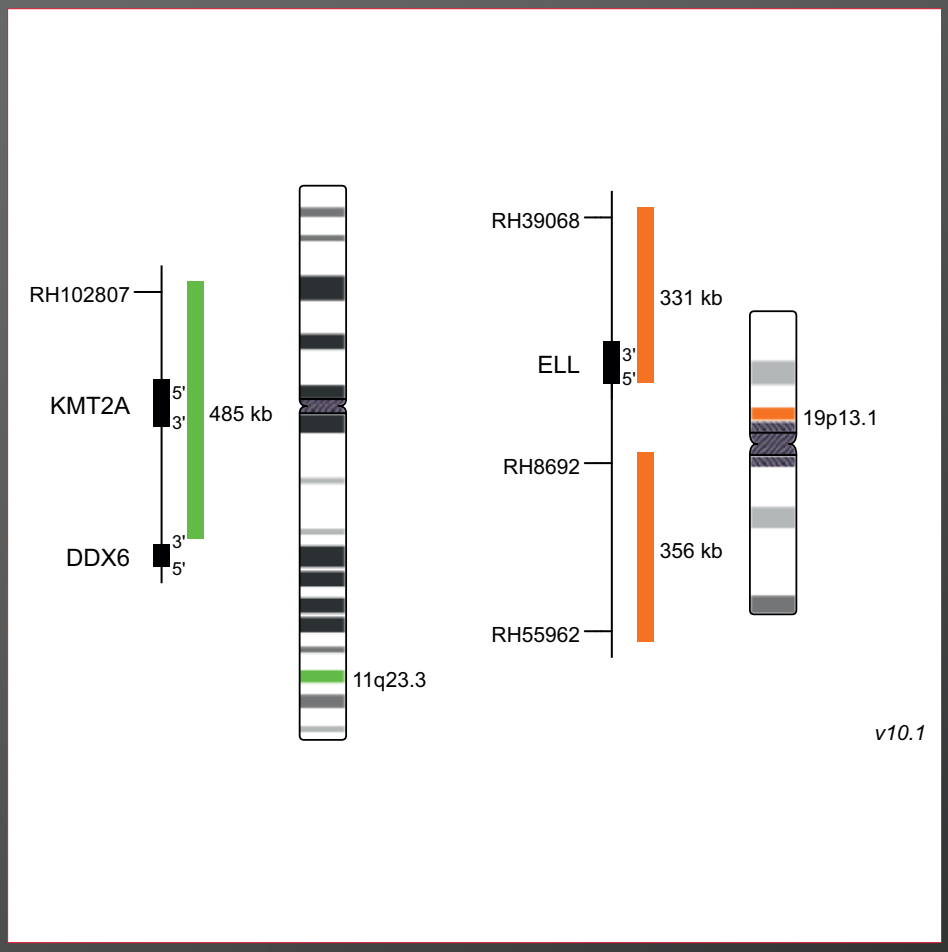
Probes



Normal Cell: Two green (2G) and two orange (2O) signals.	
Aberrant Cell (typical result): 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;p13.3) with fusion of KMT2A and MLLT1, one orange and one green signal may appear fused on metaphase spreads due to the close proximity of ELL and MLLT1.

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