XL KMT2A BA

Break Apart Probe

Order No.:
D-5090-100-OG

Description

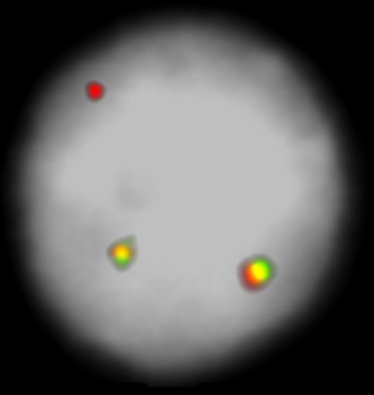
XL KMT2A BA is designed as a break apart probe. The orange labeled probe hybridizes proximal to KMT2A at 11q23 and extends into the gene up to intron 24, the green labeled probe hybridizes distal to KMT2A and extends into the gene up to intron 20 and thus overlapping each other for 3.4kb (GRCh37/hg19).

Clinical Details

The KMT2A (fomerly MLL) gene, located on chromosome 11q23, is rearranged in about 10% of all acute leukemia patients. Most of them suffer from acute lymphoblastic leukemia (ALL) or acute myeloid leukemia (AML), only a minority shows mixed lineage leukemia which has given the gene its original name ´MLL´. In infants, the incidence of KMT2A rearrangements in leukemia is 70-80%. KMT2A encodes a nuclear protein with methyltransferase activity and is part of multiprotein complexes involved in the regulation of target genes essential during early development and hematopoiesis. Today, more than 80 translocation partners of KMT2A have been identified. Translocations are resulting in in-frame fusions between the KMT2A part N-terminal to the break point cluster region and the respective fusion partners. The most common translocation partners in KMT2A associated leukemia, in the order of their prevalence are AFF1, MLLT3, MLLT1, MLLT10, ELL and AFDN. Fusion genes may also be the result of an insertion of genetic material including portions of KMT2A into other chromosomal locations. Some examples of fusion genes reported as a result of this mechanism are KMT2A-AFF1, KMT2A-MLLT3 and KMT2A-MLLT10.

The proven MetaSystems XL MLL plus D-5060-100-OG is designed to detect breaks in the KMT2A gene region. Featuring a new gene covering design, XL KMT2A BA D-5090-100-OG allows the detection of cryptic insertion of portions of KMT2A into other chromosomes as an added benefit, provided that the inserted DNA fragment is in the size range detectable by fluorescence microscopy.

- Soler et al (2008) Cancer Genet Cytogenet 183:53-59
- Meyer et al (2013) Leukemia 27:2165-2176
- Winters and Bernt (2017) Front. Pediatr. 5:4. doi: 10.3389/fped.2017.00004



XL KMT2A BA hybridized to bone marrow cells, one aberrant cell is shown. A cryptic insertion of KMT2A is observed generating a signal pattern of two orange-green colocalization/fusion signals and one additional orange signal.

.Clinical Applications

- ALL
- AML

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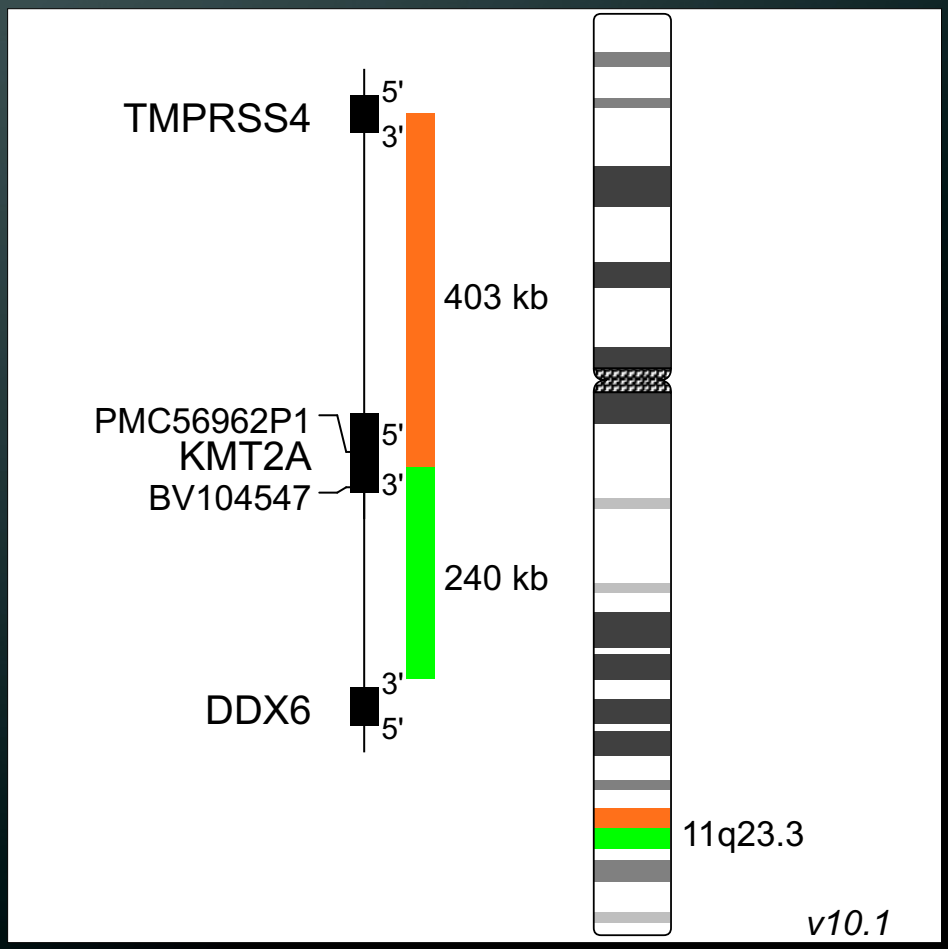
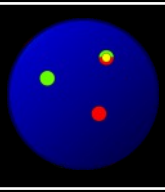
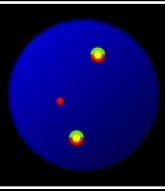


Normal cell: The expected signal pattern in normal cells is two green-orange colocalization/fusion signals (2GO).	
Typical aberrant cell: Cells with breakaparts typically have one green-orange colocalization/fusion signal plus one orange and one green signal clearly separate from one another (1G1O1GO). Breakpoints within the breakpoint cluster region result in a small orange split signal remaining with the separated green signal. The residual orange signal is significantly smaller than the separated orange signal and might even be invisible.	
Aberrant cell: An insertion of portions of KMT2A (11q23) into other chromosomes results in the signal pattern two colocalization/fusion signal plus one small clearly separated orange signal (1O2GO).	

MetaSystems Probes

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