XL ETV6 BA

Break Apart Probe

Order No.:
D-5139-100-OG

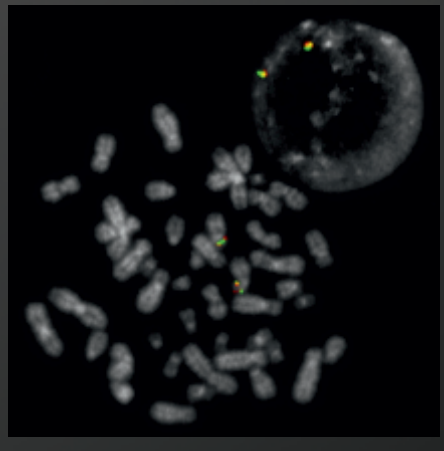
Description

XL ETV6 BA consists of an orange-labeled probe hybridizing proximal to the ETV6 gene region at 12p13.2 and a green-labeled probe hybridizing distal to the ETV6 gene region at 12p13.2 extending into the 5´gene region.

Clinical Details

The ETV6 gene (ETS variant gene 6) located on 12p13.2 codes for a transcription factor which is involved in a variety of rearrangements. Several potential mechanisms of ETV6-mediated leukemogenesis are discussed including deletions and translocations. Numerous translocations and partner genes have been identified so far. The mechanisms well described are fusion to a tyrosine kinase resulting in a constitutively active tyrosine kinase and fusions to transcription factors driving aberrant expression of target genes. The frequent translocation t(12;21)(p13;q22) results in the formation of the chimeric transcription factor ETV6::RUNX1 which can be identified in about 25% of childhood B-cell acute lymphoblastic leukemia (B-ALL) cases. The t(5;12)(q33;p13) translocation fuses ETV6 to the receptor tyrosine kinase PDGFRB (ETV6::PDGFRB). The t(9;12)(q34;p13) ETV6::ABL1 has been included in the 5th edition of the WHO Classification of Haematolymphoid Tumours in the disease type myeloid/lymphoid neoplasms with eosinophilia and tyrosine kinase gene fusions (MLN-TK). The study by Haferlach et al confirms the variety of ETV6 rearrangements in acute myeloid leukemia (AML), myelodysplastic neoplasms (MDS), and MPNs, which have been shown to be associated with other genetic events. The recurrent translocation t(7;12)(q36;p13) can be identified in pediatric acute myeloid leukemia (AML) and acute lymphoblastic leukemia (ALL) and is resulting in the fusion gene MNX1::ETV6.

XL ETV6 BA is a further developed version of XL ETV6 (D-5073-100-OG), featuring a new, partially ETV6 gene covering design, extending into the 5´-region of the ETV6 gene.



XL ETV6 BA hybridized to lymphocytes. One normal interphase and one normal metaphase are shown.

Clinical Applications

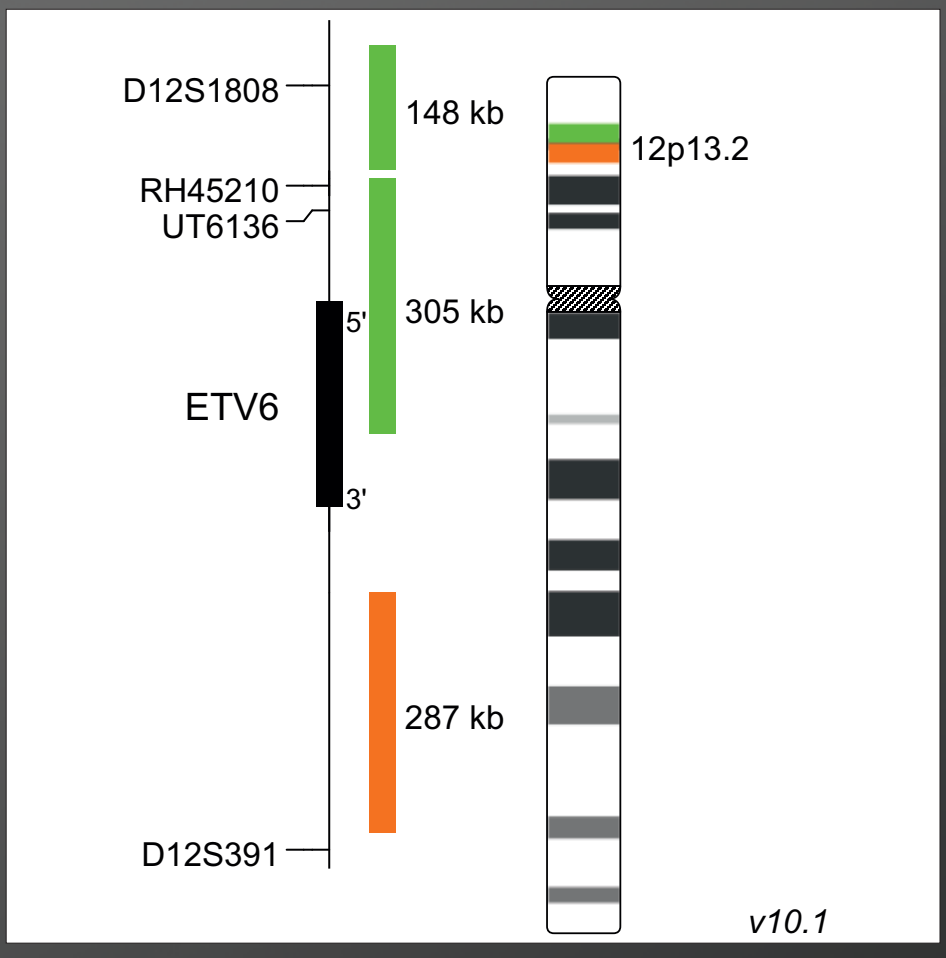
- ALL
- AML
- (CML/MPN)

Literature

- Haferlach et al (2012) Genes Chromosomes Cancer 51(4):328-337
- De Braekeleer et al (2012) Leukemia Research 36:945-961
- Naiel et al (2013) Cancers 5:281-295
- Khoury et al (2022) Leukemia 36:1703–1719

FACTSHEET

MetaSystems
Probes



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Normal cell: Two green-orange colocalization/fusion signals (2GO).	
Aberrant Cell (typical results): One green-orange colocalization/fusion signal (1GO), one separate green (1G) and orange (1O) signal each resulting from a chromosome break in the respective locus.	

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