XL P2RY8 del

Deletion Probe

Order No.:
D-5150-100-OG

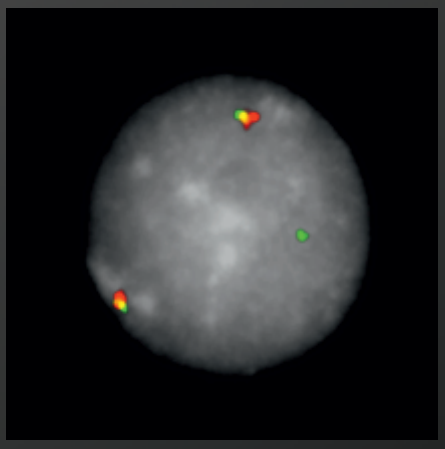
Description

XL P2RY8 del detects deletions in the short arm of chromosome X and Y at Xp22.33 and Yp11.32, respectively. The orange labeled probe spans P2RY8 and extends distally, the green labeled probe covers the 5´ end of P2RY8 and extends proximally.

Clinical Details

Acute lymphoblastic leukemia (ALL) is the most common malignancy in children (prevalence of approximately 1:1500). Children with Down syndrome have a 10- to 20-fold increased risk of developing acute leukemia. B-Cell dependent BCR-ABL1-like ALL, also known as Philadelphia chromosome (Ph)-like ALL, is a high-risk subset with a gene expression profile that shares significant overlap with that of Ph-positive (Ph+) ALL, but lacking the BCR-ABL1 fusion. In 2017, the WHO recognized BCR-ABL1-like ALL as new entity. Chromosomal rearrangements resulting in the overexpression of cytokine receptor like factor 2 (CRLF2) can be found in up to 50% of BCR-ABL1-like ALL cases. The CRLF2 gene is located in the pseudoautosomal region 1 (PAR1) of the X and the Y chromosome. CRLF2 rearrangements result in increased protein levels, which initiate significantly enhanced JAK/STAT signaling, whereby disproportionate JAK and subsequent STAT5 activation induces strongly enhanced B-cell activation and proliferation. One of the genetic mechanisms leading to constitutive overexpression of CRLF2 is a gene fusion of CRLF2 with another PAR1 gene, purinergic receptor P2Y8 (P2RY8). The resulting P2RY8-CRLF2 fusion being under the control of the P2RY8 promoter is strongly transcribed in lymphoid cells.

XL P2RY8 del can be used to detect the presence of the P2RY8-CRLF2 fusion gene.



XL P2RY8 del hybridized to bone marrow cells. One aberrant cell of a patient with a gonosomal constellation of XXY is shown. The two orange-green fusion signals represent the two unaffected CRLF2-P2RY8 loci. A deletion between CRLF2 and P2RY8 is identified by a separate green signal. This signal pattern gives strong indication that the P2RY8-CRLF2 gene-fusion is present.

Clinical Applications

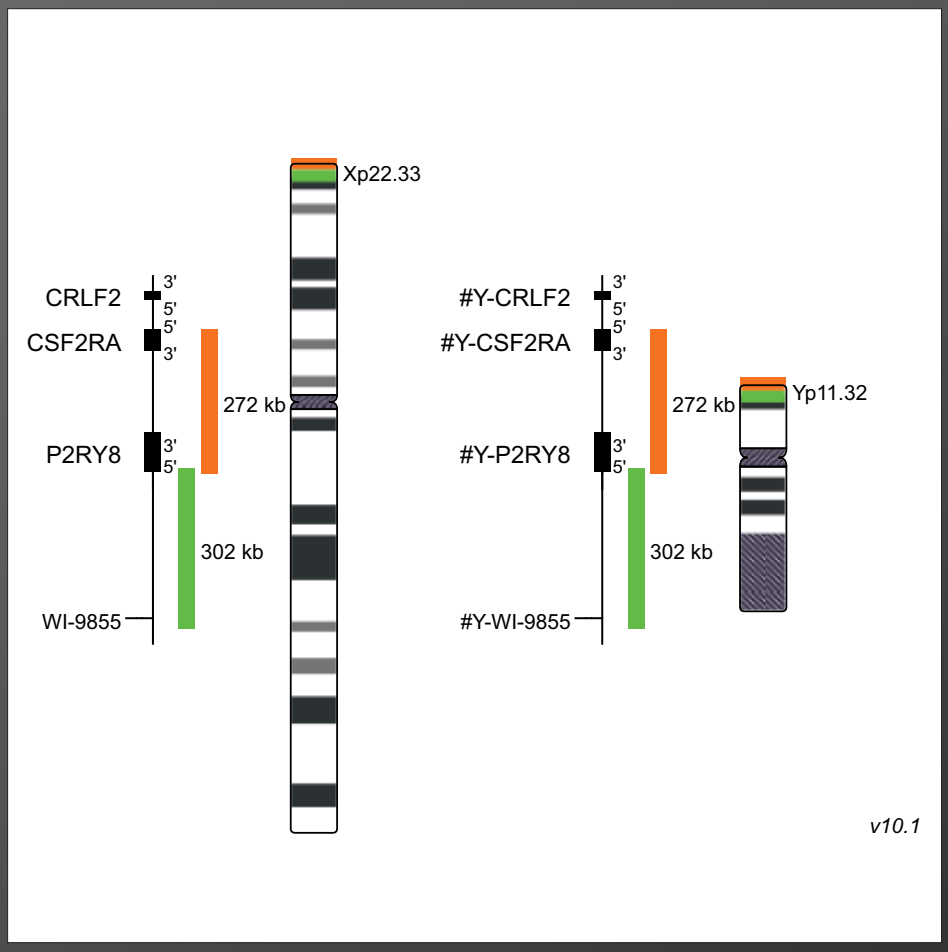
- ALL

Literature

- Mullighan et al (2009) Nat Genet 41:1243-1246
- Russell et al (2017) Genes Chromosomes Cancer 56:363-372
- Tasian et al (2017) Blood 130:2064-2072

FACTSHEET

MetaSystems Probes



Normal cell: Two green-orange colocalization/fusion signals (2GO).	
Aberrant Cell (typical results): One green-orange (1GO) colocalization/fusion signal and one green (1G) signal resulting from the loss of one orange signal.	

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