XL SPI1 BA

Break Apart Probe

Order No.:
D-5145-100-OG

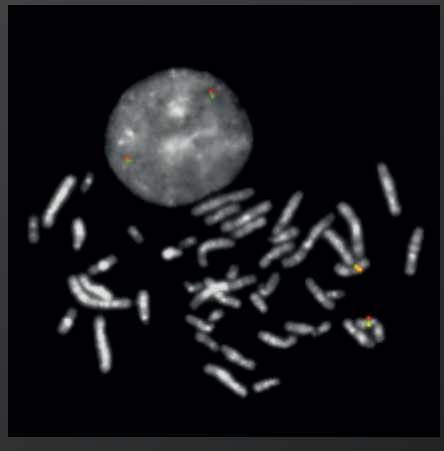
Description

XL SPI1 BA consists of an orange-labeled probe hybridizing proximal to the SPI1 gene region at 11p11.2 and a green-labeled probe hybridizing distal to the SPI1 gene region at 11p11.2.

Clinical Details

The SPI1 (SFFV provirus integration site-1) gene encodes the ETS-family transcription factor PU.1, which mediates gene expression during normal development of hematopoietic stem cells. SPI1 gene fusions were detected in 7 of 181 (3.9%) pediatric T-cell acute lymphoblastic leukemia (T-ALL) cases, analyzed and described by Seki et al. Genetic consequences of chromosomal rearrangements involving SPI1 are gene fusions containing 3' exons of SPI1 and 5' portions of TCF7, STMN1 and BCL11B. The c-terminal DNA binding domain (ETS domain) of the PU.1 protein is present in all known fusions, irrespective of the fusion partner. Fusion-positive samples show strongly increased SPI1 expression levels, as the rearranged portion of SPI1 is placed under the control of heterologous promoters of the above-mentioned genes. As SPI1 is normally expressed as an early phase gene in T-cell development, expression of wildtype SPI1 or SPI1 fusion genes at later stages results in higher cell proliferation and differentiation/maturation blockade during T-cell development. Fusion-positive cases show significantly shorter overall survival and are incurable when treated with standard chemotherapy.

SPI1 has recently been found to interact with the PML-RARα complex. Additionally, SPI1 itself is transcriptionally regulated by the PML-RARα fusion protein.



XL SPI1 BA hybridized to lymphocytes. One normal interphase and one normal metaphase are shown.

Clinical Applications

- ALL

Literature

- Martens and Stunnenberg (2010) FEBS Letters 584:2662-2669
- Seki et al (2017) Nat Genetics 49:1274-1281
- Takei and Kobayashi (2019) Int J Hematol 109:28–34

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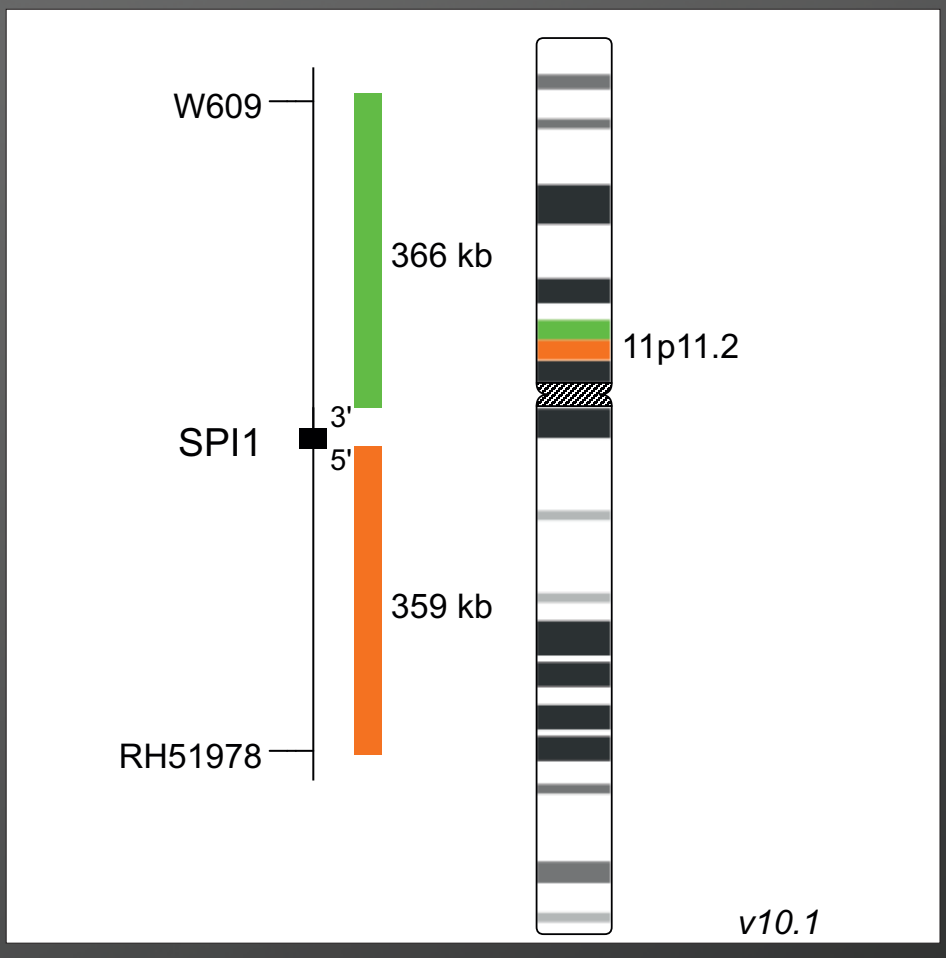

Normal cell: Two green-orange colocalization/fusion signals (2GO).	
Aberrant Cell (typical results): One green-orange colocalization/fusion signal (1GO), one separate green (1G) and orange (1O) signal each resulting from a chromosome break in the respective locus.	

MetaSystems Probes

EUROPE

Germany, Altlussheim
info@metasystems-probes.com

Italy, Milano
info@metasystems-italy.com

AMERICAS

USA, Medford
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

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MetaSystems Probes

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