XL t(5;11) NSD1/NUP98 DF

Translocation/
Dual Fusion Probe

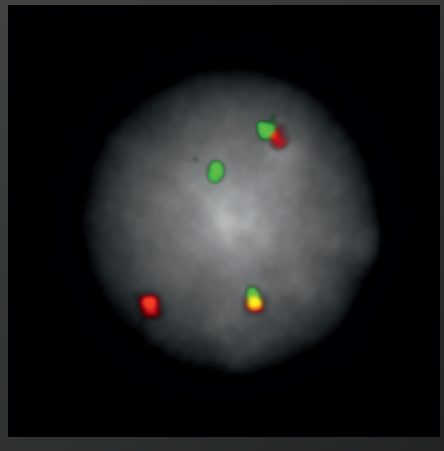
Order No.:
D-5141-100-OG

Description

XL t(5;11) NSD1/NUP98 DF consists of a green-labeled probe hybridizing to the NSD1 gene region at 5q35.2-35.3 and an orange-labeled probe hybridizing to the NUP98 gene region at 11p15.4.

Clinical Details

Acute myeloid leukemia (AML) is a rare, heterogenic disease whose prognosis varies widely, depending on several factors such as chromosomal abnormalities. Conventional cytogenetics can detect structural and numerical cytogenetic abnormalities in about 50% patients with AML. However, products from cryptic translocations, loss of chromosome material or certain fusion genes, such as t(5;11)(q35;p15) NUP98::NSD1, can only be reliably detected using FISH or molecular genetic approaches as RT-PCR technique. NUP98 (Nucleoporin 98) located at 11p15.4 encodes a protein of the nucleopore complex. So far, more than 30 different fusion partner genes of NUP98 have been identified in various leukemias. The leukemogenesis seems to be mediated by changes in chromatin structure and gene expression. NSD1 (nuclear receptor binding SET domain protein 1) located at 5q35.3 was shown to be the most frequent NUP98 fusion partner gene in pediatric AML. NSD1 is discussed to function as a transcriptional coactivator and also as a corepressor. The chimeric protein, resulting from the fusion between the N-terminal part of NUP98 including phenylalanine-glycine (FG) repeats and the C-terminal part of NSD1 induces AML in vivo and enhances the expression of HOXA and HOXB. The frequency of NUP98::NSD1 translocations in AML is low and age-dependent, with a higher frequency in younger ages than in adults. For both, pediatric and adult NUP98::NSD1-positive AML patients, the prognosis is poor and often associated with primary resistance to chemotherapy. An association with FLT3 -ITD (internal tandem duplications), and/or WT1 mutations is reported in NUP98::NSD1 positive cases, supporting the hypothesis of a multistep AML pathogenesis.



XL t(5;11) NSD1/NUP98 DF hybridized to bone marrow cells, one aberrant cell is shown. A translocation t(5;11) has occureed generating a signal pattern of two colocalization/fusion signals, one green and one orange signal.

Clinical Applications

- AML

Literature

- Jaju et al (2001) Blood 98:1264-1267
- Wang et al (2007) Nat Cell Biol 9:804-812
- Hollink et al (2011) Blood 118:3645-3656
- Fasan et al Leukemia (2013) 27:245–248
- Struski et al (2017) Leukemia 31:565-572

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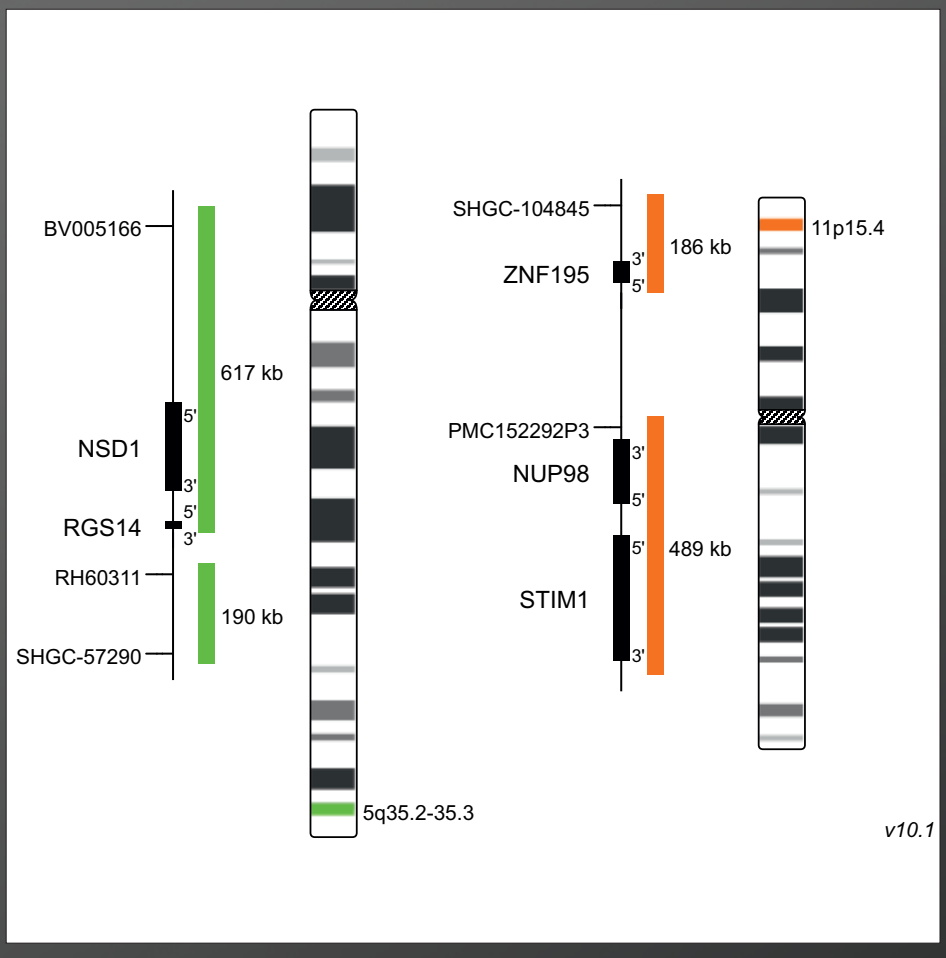

Normal Cell: Two green (2G) and two orange (2O) signals.	
Aberrant Cell (typical results): One green (1G), one orange (1O), and two green-orange colocalization/fusion signals (2GO) resulting from a reciprocal translocation between the relevant loci.	

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