# XL t(8;9) PCM1/JAK2 DF

Translocation/
Dual Fusion Probe

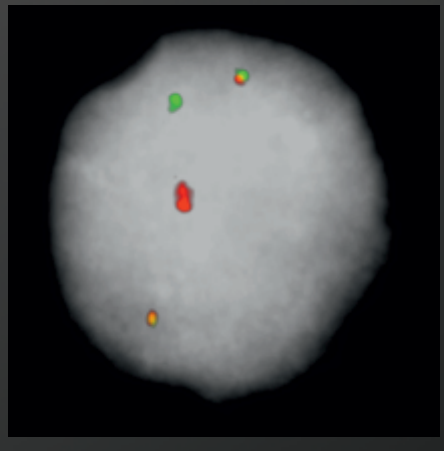
**Order No.:**
D-5120-100-OG

## Description

XL t(8;9) PCM1/JAK2 DF is designed as a dual fusion probe. The orange labeled probe spans the breakpoint at 8p22 (PCM1), and the green labeled probe spans the breakpoint at 9p24 (JAK2).

## Clinical Details

Receptor tyrosine kinases as well as Janus kinases (JAKs) have multiple functions in the signal transmission of cytokines and growth factors. Furthermore, constitutively enhanced JAK phosphorylation levels play critical roles in chronic inflammatory processes and cancer development. Chromosomal rearrangements involving JAK2, leading to constitutive activation of the kinase, have been detected in several patients with myeloproliferative neoplasm (MPN) or myelodysplastic syndrome (MDS). While ETV6-JAK2 and BCR-JAK2 fusion genes, resulting from translocations of the type t(9;12)(p24;p13) and t(9;22)(p24;q11), respectively, have been reported only in a small number of patients, pericentriolar material 1 (PCM1)-JAK2 represents the most frequent JAK2 fusion gene. The responsible translocation generating the described fusion is t(8;9)(p22;p24.1). Multiple breakpoints have been identified. In 2017 the WHO updated the category 'myeloid/lymphoid neoplasms with eosinophilia and gene rearrangement' by adding a provisional entity, PCM1-JAK2. All mentioned neoplasms described in this category are associated with rearrangements, or mutations (rarely) of four tyrosine kinases: PDGFRA, PDGFRB, FGFR1 or JAK2, respectively. As PCM1-JAK2 fusions are associated with a poor prognosis, only allogeneic hematopoietic stem cell transplantation is considered to be rather successful, although first small studies using the JAK2 inhibitor ruxolitinib as treatment for patients with PCM1-JAK2 positive chronic eosinophilic leukemia, not otherwise specified, have shown promising short-term remission.



*XL t(8;9) PCM1/JAK2 DF hybridized to bone marrow cells, one aberrant cell is shown. A translocation t(8;9) has occurred generating a signal pattern of two colocalization/fusion signals, one green and one orange signal.*

### Clinical Applications

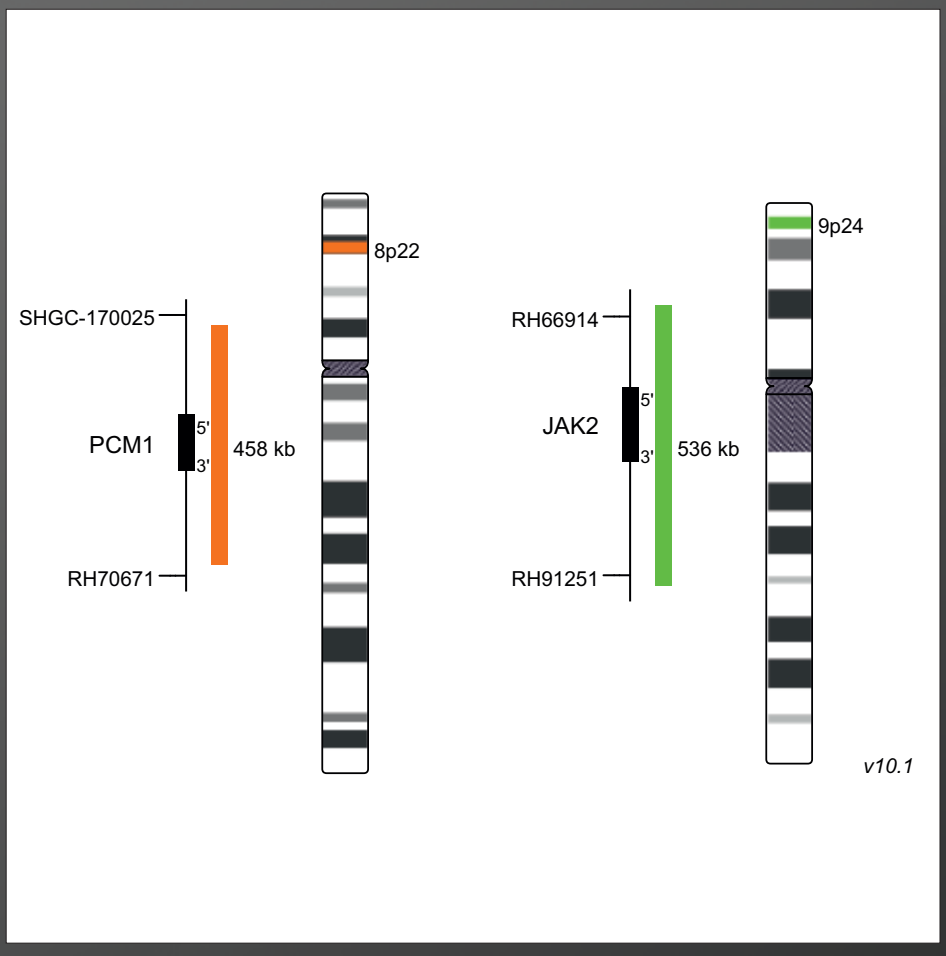
- CML/MPN

### Literature


- Lierman et al (2012) Blood 120:1529-1531
- Rumi et al (2013) J Clin Oncol 31:e269-e271
- Reiter and Gotlib (2017) Blood 129:704-714


# FACTSHEET

MetaSystems
Probes




| | |
|---|---|
| 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. |  |

# MetaSystems Probes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