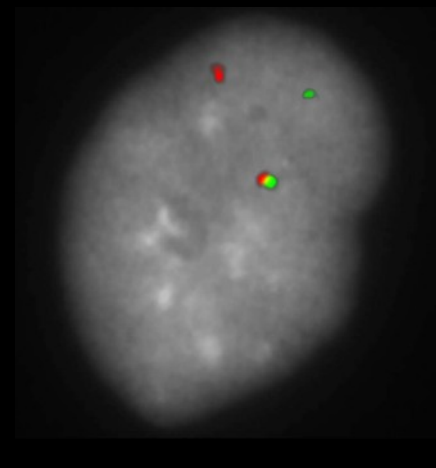# XL PDGFRA BA

Break Apart Probe

Description

XL PDGFRA BA is designed as a break apart probe. The orange labeled probe hybridizes distal to the breakpoint in the PDGFRA gene region at 4q12, the green labeled probe hybridizes proximal to the breakpoint and covers the CHIC2 gene.

Clinical Details

The category ´myeloid/lymphoid neoplasms with eosinophilia and rearrangement of PDGFRA, PDGFRB or FGFR1, or with PCM1/JAK2´ of the revised 4th edition of the WHO classification defines three particular groups with rearrangements in PDGFRA, PDGFRB or FGFR1 and a provisional entity with PCM1/JAK2 rearrangement. The clinical manifestations within this category are numerous and heterogeneous and include myeloproliferative neoplasms, myelodysplastic syndromes as well as de novo or secondary mixed phenotype acute leukemia and lymphomas. Neoplasms with eosinophilia are associated with dysregulated tyrosine kinases, usually as a result of gene fusions. It is of great importance to identify the genes involved because aberrant tyrosine kinases react with varying sensitivity to tyrosine kinase inhibitors. Patients with PDGFRA and PDGFRB rearrangements are responsive to imatinib whereas FGFR1-related diseases are non-responsive. PDGFRA rearrangements are usually associated with chronic eosinophilic leukemia (CEL). The most frequent PDGFRA-related aberration is the interstitial deletion of the CHIC2 gene with breakpoints in the FIP1L1 and PDGFRA genes. The deletion of a fragment of about 800kb is resulting in the FIP1L1-PDGFRA fusion gene, a constitutively activated tyrosine kinase transforming hematopoietic cells. In FISH assays, the detection of CHIC2 deletion at 4q12 is a surrogate for the direct detection of the FIP1L1-PDGFRA fusion gene. Translocations with other partner genes, resulting in aberrant tyrosine kinase activity, are known. Involvement of PDGFRA can be detected by break apart strategies with FISH.
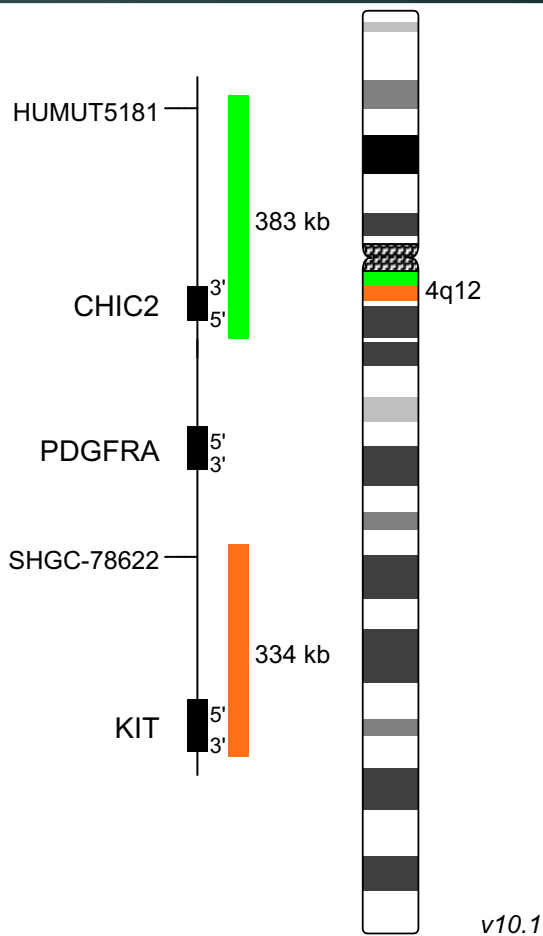

XL PDGFRA BA hybridized to bone marrow cells, one aberrant cell is shown. The expected normal signal pattern of XL PDGFRA BA is two green-orange colocalization/fusion signals representing the two normal PDGFRA loci. Cells with breakaparts typically have one normal green-orange colocalization/fusion signal plus one orange and one green signal clearly separate from one another.

Clinical Applications:

- CEL
- AML
- ALL

Literature:

- Cools et al (2003) N Engl J Med 348:1201-1214
- Pardanani et al (2003) Blood 102:3093-3096
- Swerdlow et al (2017) WHO classification of tumors of haematopoietic and lymphoid tissues (revised 4th edition)

FACTSHEET

MetaSystems
Probes

HUMUT5181 ─────

383 kb

CHIC2 ─ 3'
        5'

4q12

PDGFRA ─ 5'
          3'

SHGC-78622 ─────

334 kb

KIT ─ 5'
      3'

v10.1

Related Products

Product	Size	Order No.
XL 4q12	100µl	D-5063-100-TC
XL 4q12 DC	100µl	D-5123-100-OG

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