

## Background

The ZytoLight® SPEC PDGFRB Dual Color Break Apart Probe is designed to detect translocations involving the chromosomal region 5q32 harboring the PDGFRB gene. The PDGFRB (platelet-derived growth factor receptor-ß) gene encodes a transmembrane glycoprotein that belongs to the type III receptor tyrosine kinase family and has a key role in a variety of cellular processes.

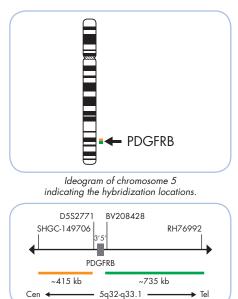
Translocations involving the PDGFRB gene are rare genetic disorders and are identified in myelodysplastic/myeloproliferative neoplasms (MDS/MPNs), chronic myeloproliferative disorders (CMPD), acute myeloid leukemia (AML), and also in atypical (BCR-ABL1-negative) chronic myeloid leukemia/chronic myelomonocytic leukemia (CML/CMML)-like diseases, often with eosinophilia and splenomegaly. The most common translocation involving PDGFRB is the t(5;12)(q32;p13.2). Result of this translocation is the fusion protein ETV6-PDGFRB, in which the pointed domain of ETV6 is juxtaposed next to the transmembrane and entire tyrosine kinase domain of PDGFRB. As a result, the tyrosine kinase is constitutively activated leading to hematopoietic cell proliferation. Patients with PDGFRB translocations respond well to imatinib therapy with excellent hematopoietic and molecular responses. Recent studies revealed that sorafenib is a further potential inhibitor of patients with ETV6-PDGFRB translocation.

**References** Bain BJ (2010) Haematologica 95: 696-8. Cross NC and Reiter A (2008) Acta Haematol 119: 199-206. Jones AV and Cross NC (2004) Cell Mol Life Sci 61: 2912-23. Keene P, et al. (1987) Br J Haematol 67: 25-31. Lierman E, et al. (2007) Haematologica 92: 27-34. Savage N, et al. (2013) Int J Lab Hematol 35: 491-500.

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## **Probe Description**

The SPEC PDGFRB Dual Color Break Apart Probe is a mixture of two direct labeled probes hybridizing to the 5q32q33.1 band. The green fluorochrome direct labeled probe hybridizes distal to the PDGFRB gene, and the orange fluorochrome direct labeled probe hybridizes proximal to the PDGFRB locus.

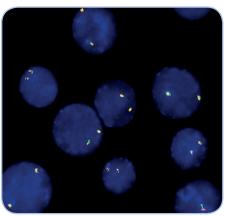


SPEC PDGFRB Probe map (not to scale).

## Results

In an interphase nucleus lacking a translocation involving the 5q32-q33.1 band, two orange/green fusion signals are expected representing two normal (nonrearranged) 5q32-q33.1 loci. A signal pattern consisting of one orange/green fusion signal, one orange signal, and a separate green signal indicates one normal 5q32-q33.1 locus and one 5q32-q33.1 locus affected by a translocation.

Molecular diagnostics simplified



SPEC PDGFRB Dual Color Break Apart Probe hybridized to normal interphase cells as indicated by two orange/green fusion signals per nucleus.

Prod. No.	Product	Label	Tests* (Volume)
Z-2197-200	Zyto <i>Light</i> SPEC PDGFRB Dual Color Break Apart Probe CE IVD	•/•	20 (200 µl)
Related Products			
Z-2028-20	Zyto Light FISH-Tissue Implementation Kit C E IVD Incl. Heat Pretreatment Solution Citric, 500 ml; Pepsin Solution, 4 ml; Wash Buffer SSC, 500 ml; 25x Wash Buffer A, 100 ml; DAPI/DuraTect-Solution, 0.8 ml		20
Z-2099-20	Zyto <i>Light</i> FISH-Cytology Implementation Kit C E IVD Incl. Cytology Pepsin Solution, 4 ml; 20x Wash Buffer TBS, 50 ml; 10x MgCl2, 50 ml; 10x PBS, 50 ml; Cytology Stringency Wash Buffer SSC, 500 ml; DAPI/DuraTect-Solution, 0.8 ml		20

\* Using 10 µl probe solution per test. CE IVD only available in certain countries. All other countries research use only! Please contact your local dealer for more information



Zyto*Light* © FISH probes are direct labeled using the unique Zyto*Light* © *Direct Label System II* providing improved signal intensity. Advanced specificity of the single copy SPEC probes is obtained by the unique ZytoVision® *Repeat Subtraction Technique*.