

# ZytoLight® SPEC SMARCB1 /22q12 Dual Color Probe

## Background

The ZytoLight® SPEC SMARCB1/22q12 Dual Color Probe is designed for the detection of deletions of the chromosomal region harboring the SMARCB1 gene. The SMARCB1 (SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1, a.k.a. INI1, SNF5, or BAF47) gene is located on chromosome 22q11.23 and encodes a tumor suppressor.

Rhabdoid tumors are highly malignant neoplasms that typically arise in infancy and early childhood. They are classified as atypical teratoid/rhabdoid tumors (AT/RT) when they occur in the CNS or as malignant rhabdoid tumors (MRT) when they are found in renal or extra-renal sites. The vast majority of AT/RTs and MRTs are characterized by loss of function of the SMARCB1 gene due to deletions or mutations. The molecular alterations are often bi-allelic resulting in complete loss of this tumor suppressor gene, and thus in cell cycle progression.

Patients with germline alterations of SMARCB1, including deletions, duplications, and mutations, were found to be predisposed to malignant rhabdoid tumors and schwannomatosis.

Moreover, deletions of the SMARCB1 gene were found to occur in patients with highly aggressive renal medullary carcinoma (RMC), epithelioid sarcoma, and poorly differentiated sarcoma.

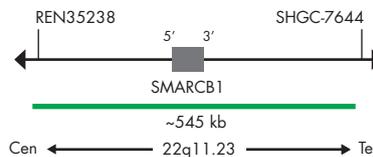
The identification of SMARCB1 deletions by FISH may represent a powerful adjunctive diagnostic tool useful in the differential diagnosis of rhabdoid tumors. Moreover, prenatal testing should be performed in situations where alterations of SMARCB1 have been documented in the family.

## Probe Description

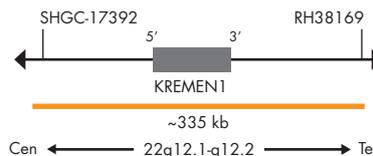
The SPEC SMARCB1/22q12 Dual Color Probe is a mixture of a green fluorochrome direct labeled SPEC SMARCB1 probe hybridizing to the human SMARCB1 gene in the chromosomal region 22q11.23 and an orange fluorochrome direct labeled SPEC 22q12 probe specific for the KREMEN1 (kringle containing transmembrane protein 1) gene region in 22q12.1-q12.2. Due to cross-hybridizations of chromosome 22 alpha satellites to other centromeric regions, probes specific for 22q12 are frequently used for chromosome 22 copy number detection.



Ideogram of chromosome 22 indicating the hybridization locations.



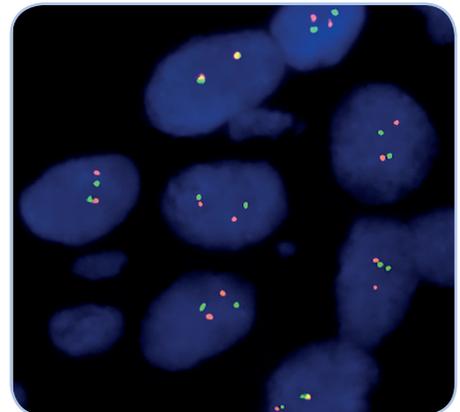
SPEC SMARCB1 Probe map (not to scale).



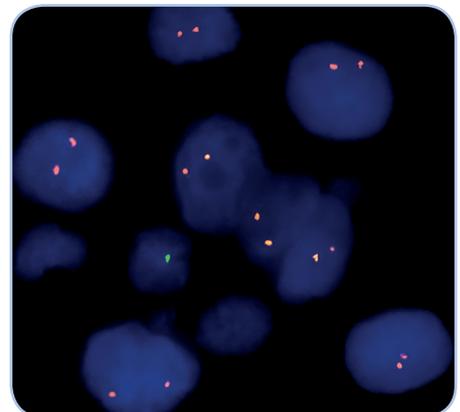
SPEC 22q12 Probe map (not to scale).

## Results

In a normal interphase nucleus, two orange and two green signals are expected. In a cell with deletion of the SMARCB1 gene locus, a reduced number of green signals will be observed. Deletions affecting only parts of the SMARCB1 gene might result in a normal signal pattern with green signals of reduced size.



SPEC SMARCB1/22q12 Dual Color Probe hybridized to normal interphase cells as indicated by two orange and two green signals per nucleus.



SPEC SMARCB1/22q12 Dual Color Probe hybridized to epithelioid sarcoma tissue section with bi-allelic deletion of the SMARCB1 gene as indicated by missing green signals in the nuclei.

## References

- Calderaro J, et al. (2012) Histopathology 61: 428-35.
- Eaton KW, et al. (2011) Pediatr Blood Cancer 56: 7-15.
- Mobley BC, et al. (2010) Acta Neuropathol 120: 745-53.
- Roberts CW & Biegel JA (2009) Cancer Biol Ther 8: 412-6.
- Sullivan LM, et al. (2013) Mod Pathol 26: 385-92.

Prod. No.	Product	Label	Tests* (Volume)
Z-2178-50	ZytoLight SPEC SMARCB1/22q12 Dual Color Probe CE IVD	●/●	5 (50 µl)

### Related Products

Z-2028-5	ZytoLight FISH-Tissue Implementation Kit CE IVD		5
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Incl. Heat Pretreatment Solution Citric, 150 ml; Pepsin Solution, 1ml; Wash Buffer SSC, 150 ml; 25x Wash Buffer A, 50 ml; DAPI/DuraTect-Solution, 0.2 ml

\* 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.