

# ZytoLight® SPEC USP6 Dual Color Break Apart Probe

## Background

The ZytoLight® SPEC USP6 Dual Color Break Apart Probe is designed to detect translocations involving the chromosomal region 17p13.2 harboring the USP6 (Ubiquitin-specific peptidase 6, a.k.a. TRE2 or TRE17) gene.

Translocations affecting USP6 have been initially found in primary aneurysmal bone cysts (ABC), a benign, but locally aggressive bone lesion that occurs predominantly during the first two decades of life. USP6 rearrangements are restricted to spindle cells in primary ABC, indistinguishable from surrounding normal spindle cells. The resulting fusion genes detected are formed by juxtaposition of the USP6 coding sequences to the highly active promoter sequences of several partner genes, as e.g. CDH11, COL1A1, OMD, TRAP150, and ZNF9, leading to the transcriptional upregulation of USP6. No true fusion genes are formed.

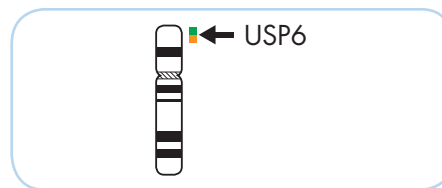
More recently, nodular fasciitis (NF), another mesenchymal lesion, has been tested positive for USP6 rearrangements. NF is a subcutaneous pseudosarcomatous myofibroblastic proliferation of unknown pathogenesis that regresses spontaneously when not surgically resected. The translocation results in the fusion of the promoter region of MYH9 located on 22q12.3 to the entire coding sequence of USP6 and subsequently in upregulated USP6 expression. For both lesions it is assumed that the detection of USP6 rearrangements by Fluorescence *in situ* Hybridization might represent a valuable diagnostic tool.

### References

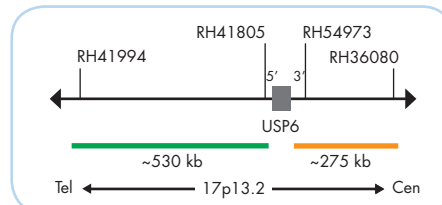
- Erickson-Johnson MR, et al. (2011) Lab Invest 91: 1427-33.
- Nakamura T, et al. (1988) Oncogene Res 2: 357-70.
- Oliveira AM, et al. (2004) Cancer Res 64: 1920-3.
- Oliveira AM, et al. (2005) Oncogene 24: 3419-26.

## Probe Description

The SPEC USP6 Dual Color Break Apart Probe is a mixture of two direct labeled probes hybridizing to the 17p13.2 band. The orange fluorochrome direct labeled probe hybridizes proximal to the USP6 gene and the green fluorochrome direct labeled probe hybridizes distal to that gene.



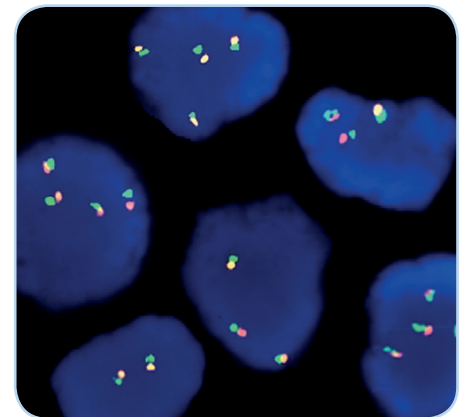
Ideogram of chromosome 17 indicating the hybridization locations.



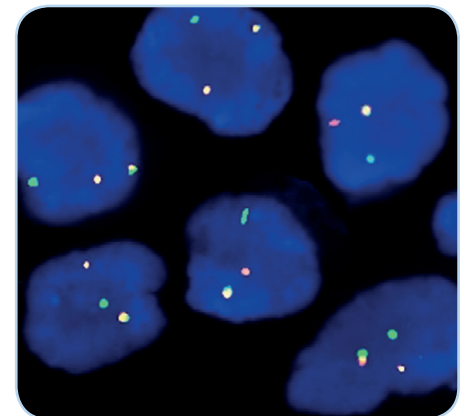
SPEC USP6 Probe map (not to scale).

## Results

In an interphase nucleus lacking a translocation involving the 17p13.2 band, two orange/green fusion signals are expected representing two normal (non-rearranged) 17p13.2 loci. A signal pattern consisting of one orange/green fusion signal, one orange signal, and a separate green signal indicates one normal 17p13.2 locus and one 17p13.2 locus affected by a translocation.



SPEC USP6 Break Apart Probe hybridized to aneurysmal bone cyst tissue section with polysomy of chromosome 17 but without translocation affecting the 17p13.2 locus as indicated by multiple orange/green fusion signals per nucleus.



Aneurysmal bone cyst tissue section with translocation affecting the 17p13.2 locus as indicated by one orange/green fusion (non-rearranged) signal, one orange signal, and one separate green signal indicating the translocation.

Prod. No.	Product	Label	Tests* (Volume)
Z-2151-50	ZytoLight SPEC USP6 Dual Color Break Apart Probe CE IVD	●/●	5 (50 µl)
<b>Related Products</b>			
Z-2028-5	ZytoLight FISH-Tissue Implementation Kit CE IVD Incl. Heat Pretreatment Solution Citric, 150 ml; Pepsin Solution, 1ml; Wash Buffer SSC, 210 ml; 25x Wash Buffer A, 50 ml; DAPI/DuraTect-Solution, 0.2 ml		5

\* 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.