Background
The ZytoLight® SPEC PDGFB Dual Color Break Apart Probe is designed for the detection of specific translocations involving the chromosomal region 22q13 harboring the PDGFB (a.k.a. SIS) gene. The PDGFB gene (platelet-derived growth factor beta polypeptide) belongs to the platelet-derived growth factor family and encodes a protein which acts as receptor tyrosine kinase. The most frequent translocation involving the PDGFB gene is t(17;22)(q22;q13) juxtaposing the PDGFB gene next to the COL1A1 gene in 17q22. Reciprocal translocations involving t(17;22)(q22;q13) are characteristic for dermatofibrosarcoma protubersans (DFSP) patients. DFSP is a highly recurrent, infiltrative skin tumor of intermediate malignancy. The rearrangements are cytogenetically characterized by the presence of supernumerary ring chromosomes containing low-level amplified sequences from chromosomes 17q22-qter and 22q10-q13.1, or unbalanced derivatives of the t(17;22) (q22;q13) translocation. The rearrangement results in a COL1A1-PDGFB fusion protein which is post-transcriptionally processed to a functional platelet-derived growth factor beta chain (PDGFB) protein. The importance of accurately diagnosing DFSP lies in its intermediate malignant nature and the availability of a therapy with significant anti-neoplastic activity but relatively minor adverse effects for cases not amenable to surgical excision.

Probes Description
The SPEC PDGFB Dual Color Break Apart Probe is a mixture of two direct labeled probes hybridizing to the 22q13 band. The orange fluorochrome direct labeled probe hybridizes proximal to the breakpoint region of the PDGFB gene, and the green fluorochrome direct labeled probe hybridizes distal to the breakpoint region of the PDGFB gene.

Results
In an interphase nucleus lacking a translocation involving the 22q13 band two orange/green fusion signals are expected representing two normal (non-rearranged) 22q13 loci. A signal pattern consisting of one orange/green fusion signal, one orange signal, and a separate green signal indicates one normal 22q13 locus and one 22q13 locus affected by a 22q13 translocation.

References