**Background**

The ZytoLight® SPEC CSF1R/D5S23,D5S721 Dual Color Probe is designed for the detection of 5q deletions. The CSF1R (colony stimulating factor 1 receptor, a.k.a. C-FMS) gene is located in the chromosomal region 5q32. The interstitial deletion of chromosome 5q is a characteristic hallmark of the myelodysplastic syndrome (MDS) with isolated del(5q). The size of the deletion as well as the breakpoints are variable but a commonly deleted region (CDR) has been narrowed to the approximately 1.5 Mb interval at 5q32-q33.1 flanked by the DNA marker D5S413 and the GLRA1 gene. One candidate gene for the development of MDS in patients with 5q-syndrome is RPS14 (ribosomal protein 14), a tumor suppressor gene located in the chromosomal region 5q33.1. Haploinsufficiency (caused by hemizygous deletion) of RPS14 is the probable cause of the erythroid defect that characterizes the 5q-syndrome. Lenalidomide has been reported to overcome the pathogenic effect of 5q deletion in MDS. Despite the severe phenotype of the 5q-syndrome, it has a relatively low (10%) transformation risk to acute myeloid leukemia (AML). Therefore, FISH may be a helpful tool for diagnosis and therapy decision.

**Probe Description**

The SPEC CSF1R/D5S23,D5S721 Dual Color Probe is a mixture of a green fluorochrome direct labeled SPEC CSF1R probe hybridizing to the CSF1R gene in the chromosomal region 5q32-q33.1 and an orange fluorochrome direct labeled SPEC D5S23,D5S721 probe specific for the chromosomal region 5p15.2-p15.31.

**Results**

In a normal interphase nucleus, two orange and two green signals are expected. In a cell with deletion of the CSF1R gene locus, one or no copy of the green signal will be observed.

**References**