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

WHO Classification of Tumors of the Haematopoietic and Lymphoid Tissues (2008); 102.