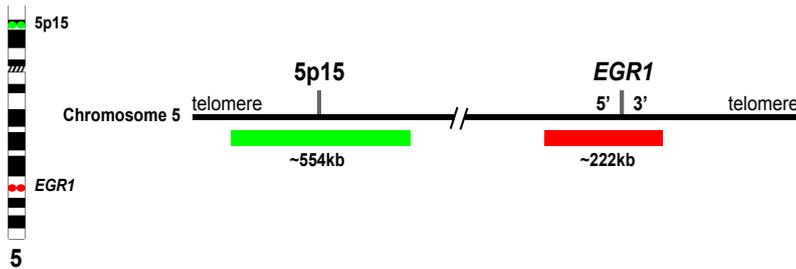


## Intended Use

The *EGR1/5p15* DNA-FISH Probe is designed to detect the deletion of the *EGR1* gene located on 5q31 relative to the control locus 5p15 by fluorescence *in situ* hybridization (FISH). The deletion of the *EGR1* gene is detected in 10-15% of *de novo* myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML) patients, and in 35-42% of therapy-related MDS (t-MDS) and therapy-related AML (t-AML) patients.<sup>[1,2]</sup> When observed as the sole chromosomal aberration in cases of MDS (also called 5q-syndrome), deletion of the *EGR1* gene is associated with a favorable prognosis and good response to lenalidomide treatment.<sup>[3]</sup> In cases of MDS/AML and t-MDS/t-AML, deletion of *EGR1* as part of a complex karyotype is associated with a worse prognosis and unfavorable outcome.<sup>[3,4]</sup>

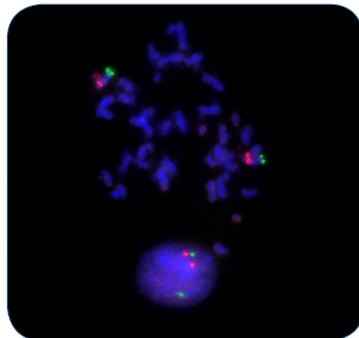


### Schematic of the *EGR1/5p15* DNA-FISH Probe:

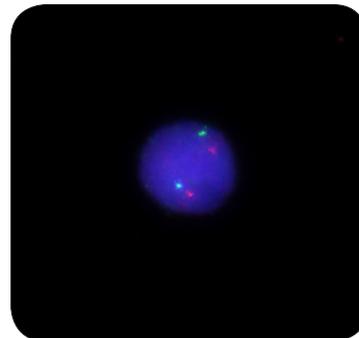
Horizontal red and green bars indicate the regions covered by the probes (approximate to scale, GRCh37/Hg19/2009). The directly labeled *EGR1* (red) probe spans the entire gene and the 5p15 (green) probe serves as a control.

## Signal Interpretation

In normal diploid metaphase and interphase nucleus, two red and two green signals would be seen corresponding to the two normal homologous chromosome 5 (Figures 1 and 2). Upon interstitial deletion of 5q31, in which the *EGR1* gene is deleted and the 5p15 band remains, one red and two green signals would be observed, which corresponds to the *EGR1* gene and two remaining 5p15 regions. Upon deletion of an entire chromosome 5, a single red and green signal would be observed, which corresponds to the remaining chromosome 5. It is recommended to confirm variant pattern or atypical signal patterns by metaphase analysis whenever possible.



**Figure 1:** Normal diploid metaphase and interphase nucleus (from normal peripheral blood specimen) with 2 red (*EGR1*) and 2 green (5p15) signals.



**Figure 2:** Normal diploid interphase nucleus (from bone marrow specimen) with 2 red (*EGR1*) and 2 green (5p15) signals.

## References

1. Herry, A., et al. *Eur J Haematol*, 2007. 78(6): p. 457-67.
2. Schoch, C., et al. *Genes Chromosomes Cancer*, 2002. 35(1): p. 20-9.
3. List, A., et al. *N Engl J Med*, 2006. 355(14): p. 1456-65.
4. Haase, D. *Ann Hematol*, 2008. 87(7): p. 515-26.

## Fluorescence Microscopy Filter Requirements

Fluorophore	Excitation <sub>max</sub>	Emission <sub>max</sub>
Green	496 nm	520 nm
Red	580 nm	603 nm
DAPI	360 nm	460 nm

Instructions for use are available at [www.cancergeneticsitalia.com](http://www.cancergeneticsitalia.com)