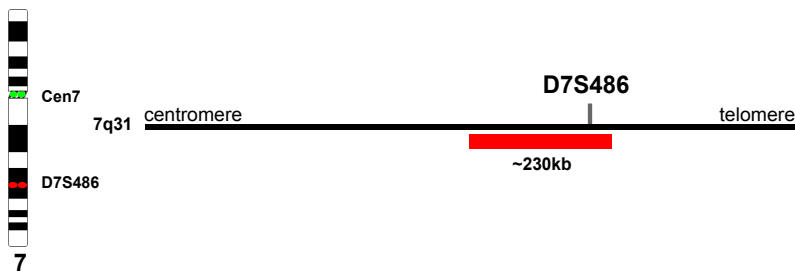


## Intended Use

The D7S486/Cen7 DNA-FISH Probe is designed to detect the deletion of the D7S486 locus located on 7q31 relative to the centromere 7 control locus, using fluorescence *in situ* hybridization (FISH). Deletions of chromosome 7 are frequently seen in many types of myeloid neoplasms. Loss of the D7S486 locus is detected in ~5% of adults with de novo myelodysplastic syndrome (MDS) and in ~50% of children with de novo MDS.<sup>[1]</sup> Additionally, loss of the D7S486 locus is observed in 5% of de novo acute myeloid leukemia (AML) patients and in 30 - 40% of therapy related MDS (t-MDS)/AML (t-AML) patients.<sup>[1,2]</sup> Deletion of the D7S486 locus or monosomy of chromosome 7 is associated with a poor prognosis in adults and children diagnosed with MDS and/or AML.<sup>[1,3]</sup>

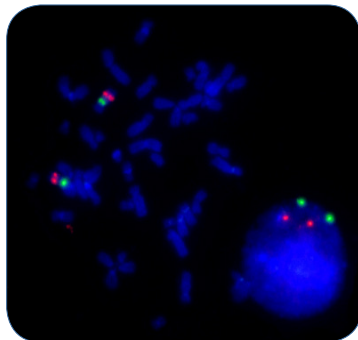


### Schematic of the D7S486/Cen7 DNA-FISH Probe:

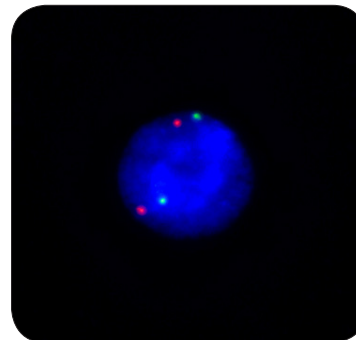
Horizontal red bar indicate the region covered by the probe (approximate to scale, GRCh37/Hg19/2009). The directly labeled D7S486 (red) probes span the D7S486 locus and the Cen7 (green) probe spans the pericentromeric region of chromosome 7.

## Signal Interpretation

In normal diploid metaphase and interphase nucleus, two red and two green signals would be observed corresponding to the normal homologous chromosome 7 (Figures 1 and 2). Upon deletion of an entire chromosome 7, a single red and green signal would be observed, which corresponds to the remaining chromosome 7. Upon interstitial deletion of 7q31, in which the D7S486 locus is deleted and the pericentromeric region remains, one red and two green signals would be observed, which corresponds to the remaining D7S486 locus and the two remaining pericentromeric regions. 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 (D7S486) and 2 green (Cen7) signals.



**Figure 2:** Normal diploid interphase nucleus (from bone marrow specimen) with 2 red (D7S486) and 2 green (Cen7) signals.

## References

1. Heim, S., Mitelman, F. (Ed). Cancer Cytogenetics, 2009 (3rd Edition). Wiley-Blackwell, New Jersey. P. 141-178.
2. Shali, W., et al., Cancer Genet Cytogenet, 2006. 168(2): p. 133-145.
3. 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)