ITALIA
Empowering Personalized Cancer Treatment

D20S108/8q11 DNA-FISH Probe

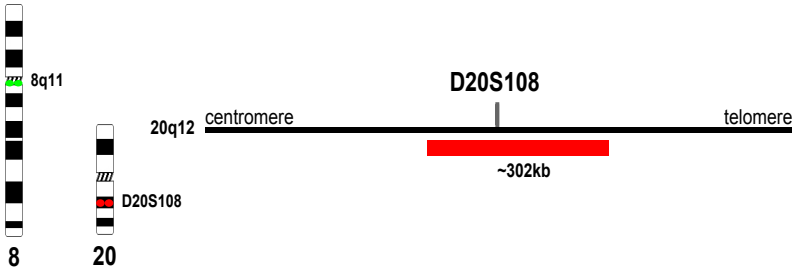
Two Color, Enumeration Probe CE

Ref: 11-001

D20S108/ 8q11

Intended Use

The D20S108/8q11 DNA-FISH Probe is designed to detect the deletion of the D20S108 locus on 20q12 and the gain of chromosome 8 using fluorescence *in situ* hybridization (FISH). Genomic copy number changes are frequent in myeloid disorders such as myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML).[1-3] Deletion of the D20S108 locus is observed in 0.6 - 5% of de novo MDS patients and in less than 2% of de novo AML patients.[1,2] In MDS patients, deletion of the D20S108 locus is associated with a good prognosis[3] whereas in AML patients it is a marker of either an intermediate or an unfavorable outcome.[2] The trisomy of chromosome 8 is observed as a sole abnormality in ~5% of MDS patients or as part of a complex karyotype in >15% of such patients and is generally associated with an intermediate prognosis.[2,3] Additionally, trisomy 8 can be observed as a sole aberration in 5% of de novo AML patients or simultaneously with other aberrations in 15% in such patients.[2] However, trisomy 8 is more common in de novo AML cases than in therapy related AML (t-AML) with an occurence of 7.4% vs. 3.3%, respectively[4,5] and has been associated with an intermediate prognosis.[2,3]



Schematic of the D20S108/8q11 DNA-FISH Probe:

Horizontal red bar indicates the region covered by the probe (approximate to scale, GRCh37/ Hg19/2009). The directly labeled D20S108 (red) probe spans the D20S108 locus and the 8q11 (green) probe spans the pericentromeric region of chromosome 8.

Signal Interpretation

In normal diploid metaphase and interphase nucleus, two green and two red signals would be observed corresponding to the two normal homologous chromosomes 8 and 20, respectively (Figures 1 and 2). Upon deletion of the D20S108 locus or deletion of an entire chromosome 20, the most commonly observed pattern is one red and two green signals, which corresponds to the remaining chromosomes 20 and 8. Trisomy 8 is detected as three green signals and is independent of whether the D20S108 locus is deleted. It is recommended to confirm variant pattern or atypical signal patterns by metaphase analysis whenever possible.

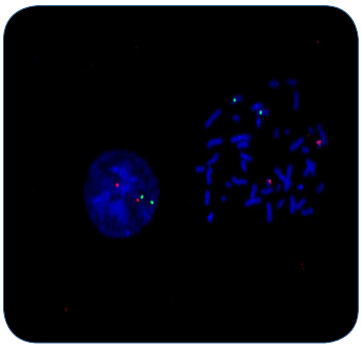


Figure 1: Normal diploid metaphase and interphase (from normal peripheral blood specimen) nucleus with 2 red and 2 green signals.

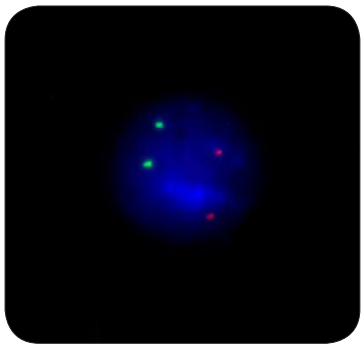


Figure 2: Normal diploid interphase nucleus (from bone marrow specimen) with 2 red and 2 green signals.

References

1. Sole, F., et al. Haematologica, 2005. 90(9):p.1168-78.
2. Heim, S., Mitelman, F. (Ed) Cancer Cytogenetics, 2009 (3rd Edition). Wiley-Blackwell, New Jersey. P. 45-178.
3. Haase, D. Ann Hematol, 2008. 87(7):p.515-26.
4. Qian, Z., et al. Chem Biol Interact, 2010. 184(1-2):p.50-7.
5. Mauritzson, N., et al. Leukemia, 2002. 16(12):p.2366-78.

Fluorescence Microscopy Filter Requirements

Fluorophore	Excitation $_{max}$	Emission $_{max}$
Green	496 nm	520 nm
Red	580 nm	603 nm
DAPI	360 nm	460 nm

Instructions for use are available at www.cancergeneticsitalia.com

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