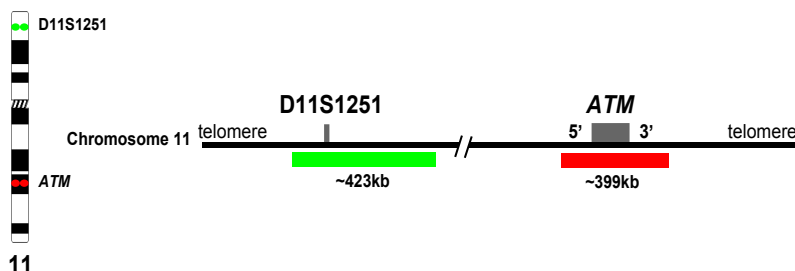


## Intended Use

The *ATM*/D11S1251 DNA-FISH Probe is designed to detect the deletion of the *ATM* gene located on 11q22 relative to the control locus D11S1251 located on 11p15 by fluorescence *in situ* hybridization (FISH). *ATM* deletions are frequently seen in several types of hematologic malignancies. The deletion of the *ATM* gene is detected in ~65% of T-cell prolymphocytic leukemia (T-PLL) cases<sup>[1]</sup>, ~50% of mantle cell lymphoma (MCL) cases<sup>[1,2]</sup>, and ~20% of chronic lymphocytic leukemia (CLL) cases.<sup>[3]</sup> Deletion of 11q in CLL patients is associated with extensive lymphadenopathy, disease progression, and shorter median survival.<sup>[3,4]</sup> Significantly improved clinical outcomes in previously untreated CLL patients with *ATM* loss have been observed using alkylating agent-based chemo-immunotherapy regimens.<sup>[5]</sup>

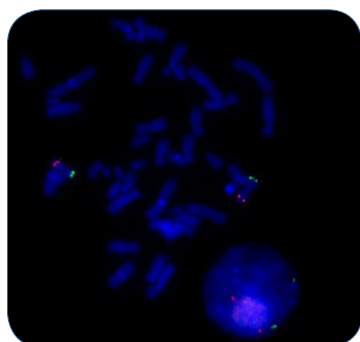


### Schematic of the *ATM*/D11S1251 DNA-FISH Probe:

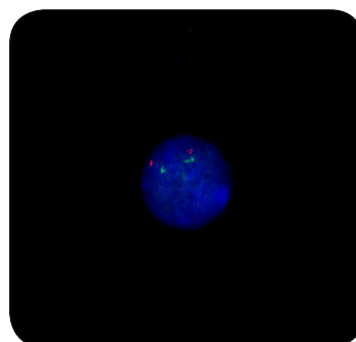
Horizontal red and green bars indicate the regions covered by the probes (approximate to scale, GRCh37/Hg19/2009). The directly labeled *ATM* (red) probe spans the entire gene and the D11S1251 (green) probe spans the locus and serves as a control.

## Signal Interpretation

In normal diploid metaphase and interphase nucleus, two red and two green signals would be observed corresponding to the two normal homologous chromosome 11 (Figures 1 and 2). Upon interstitial deletion of 11q22, in which the *ATM* gene is deleted and the D11S1251 marker remains, a single red signal and two green signals would be observed. Upon deletion of an entire chromosome 11, a single red and green signal would be observed, which corresponds to the remaining chromosome 11. 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 (*ATM*) and 2 green (D11S1251) signals.



**Figure 2:** Normal diploid interphase nucleus (from bone marrow specimen) with 2 red (*ATM*) and 2 green (D11S1251) signals.

## References

1. Monni, O., Knuutila, S. Leuk Lymphoma, 2001. 40(3-4): p. 259-66.
2. Stilgenbauer, S., et al., Blood, 1999. 94(9): p. 3262-4.
3. Dohner, H., et al. N Engl J Med, 2000. 343(26): p. 1910-6.
4. Stilgenbauer, S., et al., Leukemia, 2002. 16(6): p. 993-1007.
5. Tsimberidou, A.M., et al. Cancer, 2009. 115(2): p. 373-80.

## 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)