Leukaemia Section
Short Communication

t(12;20)(q15;q11.2)
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Clinics and pathology

Disease
Myelodysplastic/myeloproliferative syndrome, unclassifiable.

Epidemiology
Only 1 case to date: a 67 yr old female patient without BCR-ABL rearrangement.

Cytology
Basophilia (10%) and bone marrow fibrosis.

Cytogenetics
Additional anomaly: add(6).

Prognosis
Survival: 20 mths+; largely asymptomatic.

Genes involved and proteins

HGMA2
Location
12q15

Protein
Probable role in regulation of cell proliferation (transcriptional regulation of cell cycle and DNA repair genes.

Result of the chromosomal anomaly

Hybrid gene
Description
HMGA2 exon 3 spliced to intron 3 of the gene and an alternative product with exon 2 spliced to intron 2.

Fusion protein
Description
Truncated HMGA2 (normal exons 1 to 3 followed by 7 amino acids from intron 3 and normal exons 1-2plus 15 amino acids from intron 2); ectopic expression of HMGA2.

References

This article should be referenced as such: