

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

Ttruncated 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

Odero MD, Grand FH, Iqbal S, Ross F, Roman JP, Vizmanos JL, Andrieux J, Lai JL, Calasanz MJ, Cross NC. Disruption and aberrant expression of HMGA2 as a consequence of diverse chromosomal translocations in myeloid malignancies. *Leukemia*. 2005 Feb;19(2):245-52

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