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Leukaemia Section
Short Communication

t(7;12)(p12;q13)

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Clinics and pathology

Disease

Myelodysplastic syndrome (MDS)

Epidemiology

Only 1 case to date: a 33 yr old female patient with RAEB1.

Cytogenetics

Sole anomaly in this patient.

Prognosis

The patient remains asymptomatic 5 yr after presentation.

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.

Fusion protein

Description
Truncated HMGA2 (normal exons 1 to 3 followed by 12 amino acids from intron 3); ectopic expression of HMGA2.

References

Odero MD, Grand FH, Iqbal S, Ross F, Roman JP, Vizmanos JL, Andrieux J, Laï 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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