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


This article should be referenced as such: