

t(7;12)(p12;q13)

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

Gene Name [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

External links

Other database [t\(7;12\)\(p12;q13\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(7;12\)\(p12;q13\)](#) [CancerChromosomes \(NCBI\)](#)

To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:

you are welcome to submit a paper to our new [Case Report section](#).

Bibliography

Disruption and aberrant expression of HMGA2 as a consequence of diverse chromosomal translocations in myeloid malignancies.

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Leukemia. 2005; 19: 245-252.

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Contributor(s)

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