t(12;12)(p13;q13)

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Published in Atlas Database: May 2005

Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t1212p13q13ID1391.html

DOI: 10.4267/2042/38226

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Clinics and pathology

Disease
Myelodysplastic syndrome (MDS)

Epidemiology
Only 1 case to date: a 72 yr old female patient with RAEB1 which progressed to acute non lymphocytic leukemia (ANLL).

Cytogenetics
Sole anomaly in this patient.

Prognosis
The patient died 3 mths after diagnosis.

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

Fusion protein
Description
Ectopic expression of HMGA2.

References

This article should be referenced as such: