t(8;12)(p12;q15)

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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**Clinics and pathology**

**Disease**

8p11 myeloproliferative syndrome (EMS).

**Phenotype/cell stem origin**

Patients with EMS present with a myeloproliferative syndrome (MPS) with eosinophilia and a T-cell non Hodgkin lymphoma (NHL).

**Epidemiology**

Only one case to date, a 75 year old female patient (Sohal et al., 2001; Hidalgo-Curtis et al., 2008).

**Evolution**

The patient died 2 months after diagnosis, due to her lymphoma.

**Cytogenetics**

**Additional anomalies**

The t(8;12) was the sole anomaly.

**Genes involved and proteins**

**FGFR1**

**Location**

8p12

**Protein**

Receptor tyrosine kinase; contains an extracellular ligand-binding domain with Ig-like structures, a transmembrane domain, and a cytosolic tyrosine kinase (TK) domain. Involved in signal transduction.

**CPSF6**

**Location**

12q15

**Protein**

Contains a RNA recognition motif (RRM), a proline rich domain, and an arginine rich domain. Involved in pre-mRNA processing.

**Result of the chromosomal anomaly**

**Hybrid gene**

**Description**

5' CPSF6-3' FGFR1; fusion of CPSF6 intron 8 to FGFR1 exon 9, at nucleotide 1272 from ATG.

**Fusion protein**

**Description**

895 amino acids protein (97 kDa) with the RRM domain of CPSF6, fused to the TK domain of FGFR1.

**References**


Hidalgo-Curtis C, Chase A, Drachenberg M, et al. The t(1;9)(p34;q34) and t(8;12)(p11;q15) fuse pre-mRNA processing proteins SFPQ (PSF) and CPSF6 to ABL and FGFR1. Genes Chromosomes Cancer. 2008 May;47(5):379-85

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