t(1;9)(p34;q34)

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Published in Atlas Database: May 2011

Online updated version : http://AtlasGeneticsOncology.org/Anomalies/t0109p34q34ID2143.html

DOI: 10.4267/2042/46057

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**Clinics and pathology**

**Disease**

B cell progenitor acute lymphoid leukemia (B-ALL)

**Epidemiology**

Only one case to date, a 22-year-old male patient (Hidalgo-Curtis et al., 2008).

**Prognosis**

Complete remission was obtained, a relapse occurred. The patient was in complete remission 6 years after diagnosis.

**Cytogenetics**

**Cytogenetics morphological**

The translocation was found solely in the main clone, and a subclone also showed a +21.

**Genes involved and proteins**

**SFPQ**

**Location**

1p34.3

**Protein**

DNA- and RNA binding protein; pre-mRNA splicing factor; binds specifically to intronic polypyrimidine tracts.

Role in transcription and RNA splicing: SFPQ, often called PSF, is a coactivator of Fox proteins, which bind the RNA element UGCAUG and regulate alternative pre-mRNA splicing. SFPQ and NONO are part of a large complex with all the snRNPs. SFPQ is phosphorylated by GSK3, which prevents SFPQ from binding PTPRC (CD45 antigen) pre-mRNA. The association of HNRNPL and SFPQ drives the change in PTPRC (CD45) splicing (CD45 undergoes alternative splicing in response to T-cell activation).

DNA damage: DNA double-strand breaks are repaired via nonhomologous DNA end joining and homologous recombination. The SFPQ/NONO heterodimer enhances DNA strand break rejoining. SFPQ has homologous recombination and non-homologous end joining activities. SFPQ is associated with the RAD51 protein complex.

Role in transcriptional regulation: SFPQ and PTK6 (protein tyrosine kinase 6, also called BRK) play a role downstream of the EGF receptor (EGFR). SFPQ and NONO form complexes with the androgen receptor (AR) and modulate its transcriptional activity (Huret, 2011).

**ABL1**

**Location**

9q34

**Protein**

ABL1, when localized in the nucleus, induces apoptosis after DNA damage. Cyttoplasmic ABL1 has a possible function in adhesion signalling (Turhan, 2008).

**Result of the chromosomai anomaly**

**Hybrid gene**

**Description**

Break in the 3’ of SFPQ exon 10 and reunion with ABL1 intron 3; a further mRNA splicing gives rise to a chimeric SFPQ exons 1 to 9 (nucleotide 2072) fused to ABL1 exon 4 to end.
**Fusion protein**

**Description**

1609 amino acids fusion protein of 174 kDa; retains most of SFPQ, including the RNA recognition motifs and the coiled-coil domain (dimerization domain), fused to the SH2 domain of ABL1; the fusion protein also includes the SH1 domain (tyrosine kinase activity), the nuclear localization domain, and the actin binding domain of ABL1.

**Oncogenesis**

Constitutive tyrosine kinase activation is likely, through dimerization of the fusion protein.

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**References**


Hidalgo-Curtis C, Chase A, Drachenberg M, Roberts MW, Finkelstein JZ, Mould S, Oscier D, Cross NC, Grand FH.. 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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This article should be referenced as such: