t(2;5)(p21;q33)

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Clinics and pathology

**Disease**
Atypical myeloproliferative disease with eosinophilia

**Epidemiology**
One case to date, a 73-year-old female patient (Gallagher et al., 2008).

**Prognosis**
The patient was alive and well after 3 years of therapy with imatinib.

Cytogenetics

**Cytogenetics morphological**
The t(2;5) was the sole anomaly.

Genes involved and proteins

**SPTBN1**

**Location**
2p16.2 is the exact location

**Protein**
SPTBN1 (spectrin beta1 non erythrocytic), also called beta-fodrin, is a cytoskeleton protein. Forms dimers with alpha-fodrin (SPTAN1, 9q34), which self-associates head-to-head into tetramers. Membrane skeleton protein associated with ion channels and pumps (Winkelmman and Forget, 1993); Stabilizes cell surface membranes; role in mitotic spindles assembly (Bennett and Baines, 2001).

**PDGFRB**

**Location**
5q33

**Protein**
Comprises an extracellular part with 5 Ig-like C2 type domains, a transmembrane domain, and an intracellular part with a tyrosine kinase domain (made of two tyrosine kinase subdomains) for transduction of the signal. Receptor tyrosine kinase; receptor for PDGFB and PDGFD (Bergsten et al., 2001); forms homodimers, or heterodimer with PDGFRα; upon dimerization, subsequent activation occurs by autophosphorylation of the tyrosine kinase intracellular domain.

Result of the chromosomal anomaly

**Fusion protein**

**Description**
Constitutive activation of the PDGFRB tyrosine kinase domain.

References


