Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Short Communication

t(2;5)(p21;q33)

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Clinics and pathology

Disease

Atypical myeloproliferative disease with eosino-philia

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. Mem-brane skeleton protein associated with ion channels and pumps (Winkelmann 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 PDGFRA; upon dimerization, subsequent activa-tion by autophosphorylation of the tyrosine kinase intracellular domains occurs.

Result of the chromosomal anomaly

Fusion protein

Description

Constitutive activation of the PDGFRB tyrosine kinase domain.

References

Winkelmann JC, Forget BG. Erythroid and nonerythroid spectrins. Blood. 1993 Jun 15;81(12):3173-85

Bennett V, Baines AJ. Spectrin and ankyrin-based pathways: metazoan inventions for integrating cells into tissues. Physiol Rev. 2001 Jul;81(3):1353-92

Bergsten E, Uutela M, Li X, Pietras K, Ostman A, Heldin CH, Alitalo K, Eriksson U. PDGF-D is a specific, protease-activated ligand for the PDGF beta-receptor. Nat Cell Biol. 2001 May;3(5):512-6

Gallagher G, Horsman DE, Tsang P, Forrest DL. Fusion of PRKG2 and SPTBN1 to the platelet-derived growth factor receptor beta gene (PDGFRB) in imatinib-responsive atypical myeloproliferative disorders. Cancer Genet Cytogenet. 2008 Feb;181(1):46-51

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