Leukaemia Section
Short Communication

t(5;12)(q33;q24)

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Published in Atlas Database: March 2009

Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0512q33q24ID1544.html

DOI: 10.4267/2042/44692

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Clinics and pathology

Disease
Chronic myeloproliferative disease with eosinophilia

Epidemiology
Only one case to date, a 67-year-old male patient with a five years history of eosinophilia.

Prognosis
The patient was alive and well after 16 months of therapy with imatinib.

Cytogenetics

Cytogenetics morphological
The t(5;12) was the sole anomaly.

Genes involved and proteins

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 PDGF-D (Bergsten et al., 2001); forms homodimers, or heterodimer with PDGFRα; upon dimerization, subsequent activation by autophosphorylation of the tyrosine kinase intracellular domains occurs.

GIT2

Location
12q24

Protein
Numerous isoforms; the longest is made of 759 amino acids; comprises an Arf-GAP domain (amino acids 1-124), zinc-fingers (11-34), ankyrin repeats (132-161, 166-195, 199-228, according to Swiss-Prot), a Spa2-homology domain, a coiled-coil domain (leucine zipper), and a paxillin-binding site (643-679). GIT1 and GIT2 belong to the family of ADP-ribosylation factor GTPase-activating proteins (ARF-GAP). GIT1 and GIT2 form homodimers and heterodimers which bind in oligomeric complex to the p21-activated kinase-interacting exchange factor proteins ARHGEF6 and ARHGEF7, to regulate the small GTP-binding proteins RAC1 and CDC42. Associates with paxillin, and with phospholipase C (PLCG). GIT1 and GIT2 also participate in receptor internalization by regulating membrane trafficking (Hoefen and Berk, 2006).

Result of the chromosomal anomaly

Hybrid gene

Description
5' GIT2 - 3' PDGFRB. In-frame fusion between exon 12 of GIT2, and exon 11 of PDGFRB.

References


This article should be referenced as such: