t(5;12)(q33;p13) ERC1/PDGFRB

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Identity

This translocation should not be confused with the t(5;12)(q33;p13) ETV6/PDGFRB, found so far in chronic myelomonocytic leukemia (3 cases, Golub et al., 1994; Wlodarska et al., 1995; Crescenzi et al., 2007), atypical chronic myeloproliferative disorder (1 case, Nadal et al., 2006), chronic eosinophilic leukemia (1 case, Erben et al., 2010), and acute myeloid leukemia (1 case, Tokita et al., 2007). However, 26 cases of t(5;12)(q33;p13) at least have been described in the literature (see Mitelman database), most of them without molecular studies.

Clinics and pathology

Disease

Acute myeloid leukemia

Epidemiology

One case to date with ascertainment of a breakpoint in ERC1, a M5a-AML in a 36-year-old male patient.

Evolution

This leukemia was sensitive to anti tyrosine kinase treatment.

Cytogenetics

Cytogenetics morphological

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

Genes involved and proteins

PDGFRB

Location

5q32

Note

Although the translocation is described as t(5;12)(q33;p13), PDGFRB sits in 5q32.

Protein

Membrane receptor that binds specifically to PDGFB and has a tyrosine-protein kinase activity. Signal transduction.

ERC1

Location

12p13.3

Protein

Involved in exocytosis. Ubiquitously expressed. In nerve terminals, ERC1/ELKS and ERC2/CAST are localized in the active zone (AZ), a region beneath the presynaptic plasma membrane. They interact with other AZ proteins (RIM1, Piccolo, Bassoon, and Munc13-1), forming a large molecular complex, and are involved in the release of neurotransmitters (review in Hida and Ohtsuka, 2010).

Result of the chromosomal anomaly

Hybrid gene

Description

5' ERC1 - 3' PDGFRB

Transcript

ERC1 exon 15 fused to PDGFRB exon 10 (nt 3021 - nt 1837).

Fusion protein

Description

Fuses the CAZ (cytomatrix at the active zone) complex of ERC1 to the tyrosine kinase domain of PDGFRB.
References


This article should be referenced as such: