t(X;21)(q26;q22)

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Published in Atlas Database: November 2008

Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0X21q26q22ID1442.html

DOI: 10.4267/2042/44592

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Clinics and pathology

Disease
Acute myeloid leukaemia (AML)

Epidemiology
One case so far, a 56-year-old female patient with a prior history of polycythemia vera treated with hydroxyurea.

Evolution
The patient achieved complete remission, relapsed, have had a bone marrow transplantation, relapsed again and died.

Prognosis
The patient died shortly (6 months) after the discovery of the t(X;21) clone. However, it is likely that this clone developed at onset of the AML, 39 months before death.

Cytogenetics

Additional anomalies
The t(X;21) was the sole anomaly.

Genes involved and proteins

ELF4
Location: Xq25
Protein

ERG
Location: 21q22.2
Protein
Transcription factor, expressed during endothelial and hematopoietic developments.

Result of the chromosomal anomaly

Hybrid gene
Description
5’ ELF4 - 3’ ERG. The reciprocal transcript was not detected.

Fusion protein
Description
The fusion protein includes the first 25 amino acids from ELF4, encoded by exon 2, and the last 470 amino acids from ERG (exons 2 to 10). The fusion protein contains the SAM-PNT and ETS domains from ERG.

References


Moore SD, Offor O, Ferry JA, Amrein PC, Morton CC, Dal Cin P. ELF4 is fused to ERG in a case of acute myeloid leukemia with a t(X;21)(q25-26;q22). Leuk Res. 2006 Aug;30(8):1037-42

This article should be referenced as such: