Leukaemia Section
Short Communication

t(8;12)(p12;p11), ins(12;8)(p11;p12p22)

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Clinics and pathology

Disease
Non Hodgkin lymphoma (NHL) associated with myeloproliferative disease (MPD), typical of the 8p11 myeloproliferative syndrome (EMS).

Epidemiology
Only one case to date, a 75 year old male patient.

Clinics
The patient presented with a T-cell lymphoblastic Lymphoma. Complete remission was obtained on treatment, but a relapse occurred and the patient developed an acute myeloid leukaemia (AML) and died 3 months later.

Cytogenetics

Cytogenetics morphological
The patient had a ins(12;8)(p11;p12p22) at diagnosis of the NHL; monosomy 7 was found as an additioanal anomaly at the time of the AML onset

Genes involved and proteins

FGFR1
Location
8p12
Protein
FGF receptor with tyrosine kinase activity; binding of ligand induces receptor dimerization, auto-phosphorylation and signal transduction

FGFR1OP2
Location
12p12.1

Protein
Poorly known; expressed in various tissues; no significant homology to any known protein; comprises coiled-cil domains.

Result of the chromosomal anomaly

Hybrid gene
Description
5' FGFR1OP2 - 3' FGFR1; in frame fusion of exon 4 of FGFR1OP2 to exon 9 of FGFR1 (nucleotide 1817 of FGFR1OP2 intron 4 joined to nucleotide 1132 of FGFR1 intron 8). Reciprocal transcripts were not detected. The direction of transcription of FGFR1OP2 is telomere to centromere, while FGFR1's transcription is centromere to telomere, implying an inversion.

Fusion protein
Description
The first 2 coiled-coil domains of FGFR1OP2 is fused to the COOH terminal part of FGFR1, including part of its juxta membrane domain, and its tyrosine kinase domain: 132 amino acids from FGFR1OP2 and 394 from FGFR1. The FGFR1OP2-FGFR1 fusion protein may exhibit constitutive kinase activity, inducing dimerization, constitutive signal transduction, and be responsible for transforming activity.

References


This article should be referenced as such: