Abstract

Review on t(20;21)(q11;q22), with data on clinics, and the genes involved.

Keywords
Chromosome 20; Chromosome 21; Acute myeloid leukemia; Chronic myelogenous leukaemia; RUNX1. NOL4L

Clinics and pathology

Disease
Acute myeloid leukaemia (AML) without maturation (M1- AML) in one case, AML with maturation (M2-AML) in one case; and blast crisis of myeloid type of chronic myelogenous leukaemia in two cases.

Epidemiology
Four cases available to date; three with t(20;21)(q11;q22) RUNX1/? (Misawa et al. 1986; Dube et al., 1989; Jeandidier et al., 2006), and one case with proved t(20;21)(q11.2;q22.1) RUNX1/NOL4L (Guastadisegni et al. 2010). They were all male patients, aged aged 44, 57, 67, and 72 years.

Cytogenetics

Additional anomalies
No additional anomaly in one case, monosomy 5 and major karyotypic anomalies in one case,
monosomy 7 in another case, and t(9;22)(q34;q11) in the remaining case.

Genes involved and proteins

NOL4L (nucleolar protein 4 like)
Location 20q11.2
DNA/RNA
11 exons
Protein
Nothing is known concerning the domains of the protein, nor it's function.

RUNX1 (runt-related transcription factor 1 (acute myeloid leukemia 1; aml1 oncogene))
Location 21q22.12
Protein
RUNX1, also called AML1 or CBFA2, contains a Runt domain and, in the C-term, a transactivation domain, an inhibition domain, and various regulatory regions; forms heterodimers; widely expressed; nuclear localization.
RUNX1 is a transcription factor, critical regulator of hematopoietic-cell development.
It binds to the core site 5’ PyGPyGGTPy 3’ of promoters and enhancers.
RUNX1 is involved in many de novo and treatment related leukemias.
## Result of the chromosomal anomaly

### Hybrid gene

**Description**
RUNX1 exon 6 was fused to NOL4L exon 8 in the case reported by Guastadisegni et al. 2010.

### Fusion protein

**Oncogenesis**
Wild-type NOL4L was expressed at low levels in AML and normal bone marrow, whereas the RUNX1/NOL4L was expressed at high levels (Guastadisegni et al. 2010).

### References


*This article should be referenced as such:*