

Leukaemia Section

Short Communication

t(1;14)(q21;q32) MUC1/IGH

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Clinics and pathology

Note

The chromosomal band 1q21 is the third most frequent site of rearrangement in non-Hodgkin's lymphoma after 14q32 and 18q21.

Five genes mapped to this region (BCL9, MUC1, FCGR2B, MUM2, API2) some have been shown to be deregulated by juxtaposition with the IgH genes.

Disease

B-cell non Hodgkin Lymphoma (NHL).

Epidemiology

The MUC1 region is rearranged in 6% of tumors with 1q21 cytogenetic aberration.

Cytology

No clear association with a particular NHL subtype has been reported.

Prognosis

Poor prognosis especially in diffuse large cell lymphoma.

May be associated with tumor progression.

Cytogenetics

Cytogenetics morphological

A number of 1q21 abnormalities result in an unbalanced chromosome 1 translocation.

Additional anomalies

Caryotype of tencomplex. Usually detected with t(14;18)(q32;q21) and t(8;14)(q24;q32) as a secondary chromosomal abnormalitie.

Genes involved and proteins

MUC1

Location

1q21.

Note

Located 8cM telomeric to BCL9; aliases of MUC1 are EMA and CD227.

DNA/RNA

3.88 kb, 8 exons, 1721 bp, 2 transcripts.

Protein

122.1 kDa (1255 aa) Highly glycosylated protein.

The MUC1 protein can be expressed as a transmembrane or secreted protein. May be playing a role in adhesive functions and in cell-cell interactions, metastasis, signaling and is implicated in some adenocarcinomas. The EMA which is equivalent to MUC1 occurs in lymphocyte-predominant Hodgkin's disease, plasmocytomas and T-cell lymphomas due to mechanisms other than 1q21 rearrangement.

IgH

Location

14q32.

Result of the chromosomal anomaly

Hybrid gene

Description

The translocation links sequences 2.4 kpb 3' of the MUC1 gene on chromosome 1 to the IGH4 switch

region on chromosome 14. MUC1 gene is intact. The MUC1 gene is brought into proximity with the C gamma 4 and C alpha 2 loci. Downstream of C alpha 2 is an enhancer element implicated in the activation of MUC1 expression.

Fusion protein

Description

No fusion protein.

Oncogenesis

Chromosomal translocation involving class switch recombination when DNA strand breaks are introduced into the switch regions of recombining CH genes. Activation of MUC1 translation and transcription. An important role for MUC1 in tumorigenesis has been demonstrated in Muc-1 null mice. MUC1 is associated with delayed progression of the tumor (selective advantage, inhibition of cell adhesion).

In addition to activation of MUC1, haploid loss of chromosome 1 long arm also contributes to oncogenesis in some tumors.

References

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