

Leukaemia Section

Short Communication

t(2;18)(q11;q21)

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Clinics and pathology

Disease

Non Hodgkin lymphoma.

Phenotype/cell stem origin

One case to date, a 65-year-old female patient with a follicular lymphoma stage II-a (Impera et al., 2008).

Evolution

Complete remission was obtained.

Cytogenetics

Additional anomalies

A complex karyotype was found, with +11, and other anomalies.

Genes involved and proteins

AFF3

Location

2q11.2

Protein

AFF3 belongs to a family of putative transcription factors also comprising AFF1 (AF4, FEL, MLLT2) in 4q21, AFF2 (FMR2, FRAXE) in Xq28 and AFF4 (AF5Q31) in 5q31. AFF3 has been found a susceptibility gene in autoimmune diseases, namely rheumatoid arthritis, psoriatic arthritis, and juvenile idiopathic arthritis (Barton et al., 2009; Castelino and Barton, 2010; Hinks et al., 2010). AFF3 is deleted in Nievergelt syndrome, an autosomal dominant mesomelic dysplasia (Steichen-Gersdorf et al., 2008). AFF3 was also found expressed in 20% of mammary tumor cells but not in normal acini in a study (To et al., 2005).

BCL2

Location

18q21.33

Protein

Antiapoptotic protein.

Result of the chromosomal anomaly

Hybrid gene

Description

Fusion of AFF3 exon 1 to BCL2 exon 2.

Fusion protein

Oncogenesis

Leads to the overexpression of BCL2.

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