t(2;18)(q11;q21)
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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 (FM2R, 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
Fusion of AFF3 exon 1 to BCL2 exon 2.

Fusion protein

Oncogenesis
Leads to the overexpression of BCL2.

References


Castelino M, Barton A. Genetic susceptibility factors for psoriatic arthritis. Curr Opin Rheumatol. 2010 Mar;22(2):152-6


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