t(2;17)(p23;q25)

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Clinics and pathology

Disease
Anaplastic large cell lymphoma: translocations involving 2p23 are found in more than half cases of anaplastic large cell lymphoma (ALCL), a high grade non Hodgkin lymphoma (NHL). They involve ALK, and are therefore called ALK+ ALCL.

The most frequent ALK+ ALCL being the the t(2;5)(p23;q35) with NPM1-ALK fusion protein, which localises both in the cytoplasm and in the nucleus.

The t(2;17)(p23;q25) has so far been described in only 1 case, and, like other t(2;Var) involving various partners and ALK, the fusion protein has a cytoplasmic localization; they are therefore called "cytoplasm only" ALK+ ALCL.

Clinics
ALK+ ALCL without the t(2;5) (so called cytoplasmic only ALK cases) show clinical features similar to those of classical ALK+ ALCL: young age, male predominance, presentation with advanced disease, systemic symptoms, frequent involvement of extranodal sites, and a good prognosis. The t(2;17) case was that of a 53 yrs old man.

Genes involved and proteins

ALK
Location
2p23

Protein
1620 amino acids; 177 kDa; glycoprotein (200 kDa mature protein); membrane associated tyrosine kinase receptor.

ALO17 (ALK lymphoma oligomerization partner on chromosome 17)
Location
17q25

Result of the chromosomal anomaly

Hybrid gene
Description
5' ALO17 - 3' ALK

Fusion protein
Description
N term ALO17 fused to the 562 C-term amino acids from ALK (i.e. the entire cytoplasmic portion of ALK with the tyrosine kinase domain).

Expression / Localisation
Cytoplasmic localisation (in contrast with the t(2;5)(p23;q35) with NPM1-ALK, which localizes both in the cytoplasm and in the nucleus).

References


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