t(1;2)(q25;p23)

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

The t(1;2)(q25;p23) is very rare, 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.

**Epidemiology**

A very few (four) cases known so far.

**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. Nothing in particular is known concerning t(1;2) cases, as cases are not documented.

**Cytogenetics**

Complex karyotypes and/or hidden translocation in the 2 cases with cytogenetic data; FISH analyses are essential.

**Disease**

Inflammatory myofibroblastic tumors.

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### Genes involved and proteins

**TPM3 (tropomyosin alpha chain)**

**Location**

1q25

**Protein**

284 amino acids, 33 kDa; coiled coil structure; role in Calcium dependant actin-myosin interaction.

**ALK**

**Location**

2p23

**Protein**

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

**Result of the chromosomal anomaly**

**Hybrid gene**

**Description**

5’ TPM3 - 3’ ALK.
**Fusion protein**

**Description**
104 kDa ; 221 (?) N-term amino acids from TPM3 fused to the 562 C-term amino acids from ALK (i.e. the entire cytoplasmic portion of ALK with the tyrosine kinase domain); homodimerization of the fusion protein.

**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).

**Oncogenesis**
TPM3-ALK is continuously activated.

**References**

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