t(2;3)(p23;q21)

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Identity

2 3

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G- banding - Courtesy Melanie Zenger and Claudia Haferlach.

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;3)(p23;q21) 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

Only 2 cases to date: a 10 yr old girl and a 19 yr old man.

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(2;3) cases.

Genes involved and proteins

ALK

Location
2p23

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

TFG (tropomyosin receptor kinase-fused gene)

Location
3q21

Protein
406 amino acids, 44 kDa; widely expressed.

Somatic mutations

Apart from the TFG-ALK herein described (see below), TFG is also known to de fused to NTRK1 in a subset of thyroid papillary carcinomas.

Result of the chromosomal anomaly

Hybrid gene

Description
5’ TFG - 3’ ALK.
**Fusion protein**

**Description**
83 kDa and 96-97 kDa; as the breakpoint in TPM3 was variable, 701 amino acids in the sort er fusion protein, composed of 138 N-term amino acids from TFG, including the ciolet-coil oligomerization domain 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).

**References**


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This article should be referenced as such: