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Leukaemia Section
Short Communication

t(2;17)(p23;q23)

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Clinics and pathology

Disease

Anaplasic large cell lymphoma: translocations involving 2p23 are found in more than half cases of anaplasic 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;q23) 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 14 yrs old girl.

Genes involved and proteins

ALK

Location
2p23

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

CLTC

Location
17q23

Protein
1675 amino acids, 191 kDa; component of the vesicles matrix originated from the plasma membrane or the Golgi.

Result of the chromosomal anomaly

Hybrid gene

Description
5' CLTC - 3' ALK

Fusion protein

Description
The 1634 N term amino acids from CLTC 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).

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