

Leukaemia Section

Short Communication

t(X;2)(q11;p23)

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Clinics and pathology

Disease

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.

In translocations other than the t(2;5), i.e. in t(2;Var) involving various partners and ALK, the fusion protein has a cytoplasmic localization; they are therefore called "cytoplasm only" ALK+ ALCL. However, in case of a t(X;2) the localization is restricted to the membrane. The t(X;22)(q11;p23) is very rare.

Phenotype/cell stem origin

CD30+; ALK+.

Clinics

Only 1 case to date: a 18 yr old man.

Prognosis

Unknown: the patient achieved remission, but died of an unrelated cause.

Genes involved and proteins

MSN

Location

Xq11

Protein

576 amino acids, 75 kDa; cytoskeleton protein; binds to the plasma membrane and interacts with actin.

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' MSN - 3' ALK. The breakpoint in ALK is different (17 bp downstream) from that observed in NPM1-ALK and other hybrid genes.

Fusion protein

Description

1005 amino acids, 125 kDa; 448 N-term amino acid from MSN, containing the band 4.1 like domain and most of the alpha helix domain, fused to the 557 (instead of the usual 562) C-term amino acids from ALK (i.e. the cytoplasmic portion of ALK with the tyrosine kinase domain).

Expression / Localisation

Membrane restricted, in contrast with the t(2;5)(p23;q35) with NPM1-ALK, which localizes both in the cytoplasm and in the nucleus, and with other "variant ALK+" which have a cytoplasmic localisation.

Oncogenesis

Tyrosine kinase activity.

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