

Leukaemia Section

Short Communication

t(2;22)(p23;q11.2)

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Published in Atlas Database: August 2003

Online updated version: <http://AtlasGeneticsOncology.org/Anomalies/t0222p23q11D1291.html>
DOI: 10.4267/2042/38025

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Clinics and pathology

Disease

Anaplastic large cell lymphoma (ALCL): 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;22)(p23;q11) 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;22) case was that of a 12 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.

MYH9

Location

22q11.2

Protein

Codes for the non-muscle myosin II, heavy chain type A. Non-muscle myosin II is involved in motility and cell division.

Germinal mutations

In patients with May-Hegglin anomaly and Fechner syndrome.

Somatic mutations

MYH9/ALK fusion protein in ALCL (see above).

Result of the chromosomal anomaly

Hybrid gene

Description

5' MYH9 - 3' ALK

Fusion protein

Description

2201 amino acids, 220 kDa. N term MYH9 fused to the 556 C-term amino acids from ALK (i.e. the entire cytoplasmic portion of ALK with the tyrosine kinase domain), instead of the classic 562 C-term amino acids from ALK seen in other ALK fusion proteins.

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

Drexler HG, Gignac SM, von Wasielewski R, Werner M, Dirks WG. Pathobiology of NPM-ALK and variant fusion genes in

anaplastic large cell lymphoma and other lymphomas. *Leukemia*. 2000 Sep;14(9):1533-59

Stein H, Foss HD, Dürkop H, Marafioti T, Delsol G, Pulford K, Pileri S, Falini B. CD30(+) anaplastic large cell lymphoma: a review of its histopathologic, genetic, and clinical features. *Blood*. 2000 Dec 1;96(12):3681-95

Delsol G, Ralfkiaer E, Stein H, Wright D, Jaffe E. Anaplastic large cell lymphomas, Primary systemic (T/Null cell type). *WHO Classification of Tumors. Pathology and Genetics of tumours of Haematopoietic and Lymphoid Tissues* . 2001 pp 230-235.

Morris SW, Xue L, Ma Z, Kinney MC. Alk+ CD30+ lymphomas: a distinct molecular genetic subtype of non-Hodgkin's lymphoma. *Br J Haematol*. 2001 May;113(2):275-95

Lamant L, Gascoyne RD, Duplantier MM, Armstrong F, Raghav A, Chhanabhai M, Rajcan-Separovic E, Raghav J, Delsol G, Espinos E. Non-muscle myosin heavy chain (MYH9): a new partner fused to ALK in anaplastic large cell lymphoma. *Genes Chromosomes Cancer*. 2003 Aug;37(4):427-32

This article should be referenced as such:

Huret JL. t(2;22)(p23;q11.2). *Atlas Genet Cytogenet Oncol Haematol*. 2003; 7(4):272-273.
