Leukaemia Section

Short Communication

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

Jean-Loup Huret

Genetics, Dept Medical Information, UMR 8125 CNRS, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

Published in Atlas Database: August 2003

Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0222p23q11D1291.html

DOI: 10.4267/2042/38025

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.

© 2003 Atlas of Genetics and Cytogenetics in Oncology and Haematology

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

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


This article should be referenced as such: