

# Leukaemia Section

## Short Communication

### t(2;3)(p23;q21)

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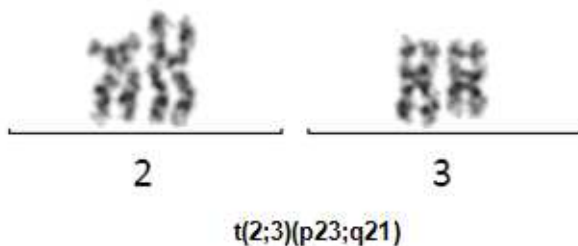
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#### Identity



t(2;3)(p23;q21) 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 be 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 coiled-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**

Hernández L, Pinyol M, Hernández S, Beà S, Pulford K, Rosenwald A, Lamant L, Falini B, Ott G, Mason DY, Delsol G, Campo E. TRK-fused gene (TFG) is a new partner of ALK in anaplastic large cell lymphoma producing two structurally different TFG-ALK translocations. *Blood*. 1999 Nov 1;94(9):3265-8

Rosenwald A, Ott G, Pulford K, Katzenberger T, Kühl J, Kalla J, Ott MM, Mason DY, Müller-Hermelink HK. t(1;2)(q21;p23) and t(2;3)(p23;q21): two novel variant translocations of the t(2;5)(p23;q35) in anaplastic large cell lymphoma. *Blood*. 1999 Jul 1;94(1):362-4

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

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*This article should be referenced as such:*

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