

Leukaemia Section

Mini Review

t(9;22)(p24;q11.2)

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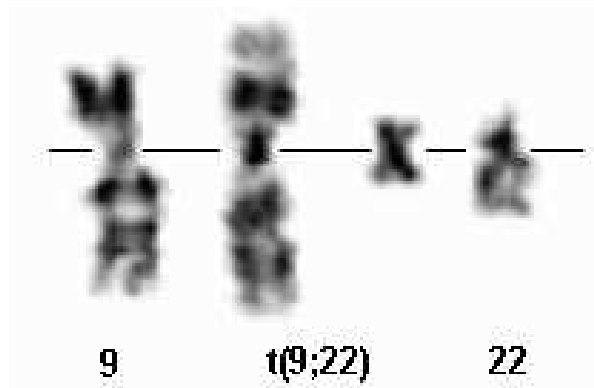
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Identity

Note: Only one case with this translocation has been reported yet.



G-banded chromosomes showing t(9;22)(p24;q11.2).

Clinics and pathology

Disease

Typical chronic myeloid leukemia (CML).

Phenotype / cell stem origin

Hematopoietic stem cell?

Epidemiology

Only one case described so far.

Treatment

No response to Imatinib!

Prognosis

Blast crisis developed 20 months after initial diagnosis.

The patient died 24 months after initial.

Cytogenetics

Cytogenetics molecular

FISH with a BCR/ABL probe (dual color dual fusion) will show a split of the BCR signal but no fusion signals and two normal ABL signals.

Additional anomalies

7q deletion and trisomy 19 was found at blast crisis.

Genes involved and Proteins

BCR1

Location: 22q11.2

JAK2

Location: 9p24

Protein

Janus activated kinase 2, protein tyrosine kinase.

Results of the chromosomal anomaly

Hybrid gene

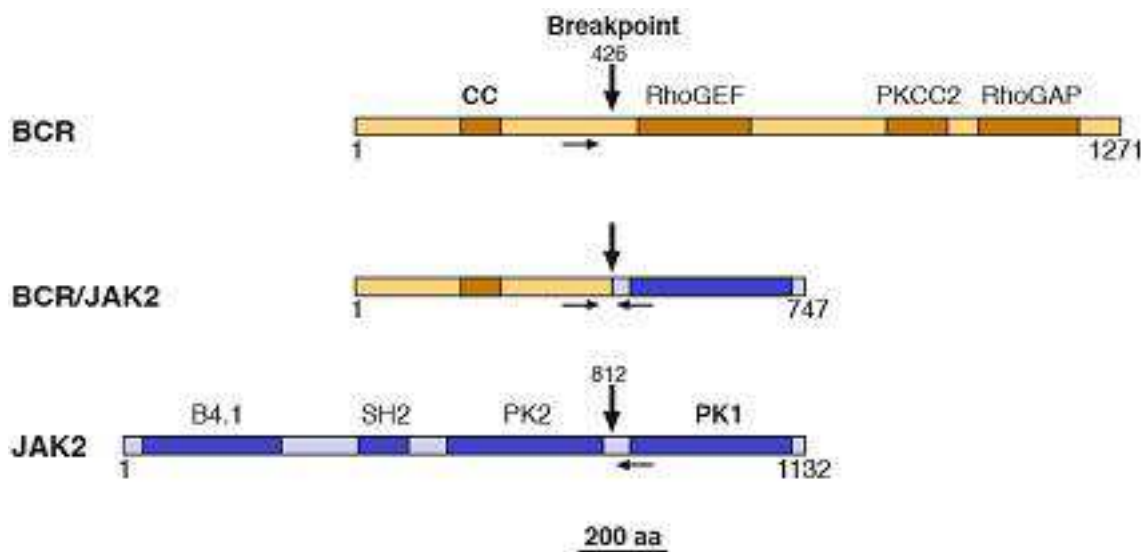
Note: Only the BCR-JAK2 fusion transcript was detected. The reciprocal JAK2-BCR fusion transcript could not be amplified.

Detection protocole

The fusion transcript can be detected by RT-PCR using the 5' BCR sense primer: 5'-cagaactcgaacagtccttc-3' (bp 1602-1622) and the 3' JAK2 antisense primer: 5'tcataccggcacatctccacac-3' (bp 3100-3081). A PCR product of 300 bp should be expected. Please note that since only one case is known, the breakpoints may vary slightly in future cases. This might necessitate the design of different primers.

BCR	nt	1770
	CAT GGA GAC GCA GAT GGC TCG TTC	
	H G D A D G S F	
	aa	427
BCR/JAK2	CAT GGA GAC GCA Gat tat gaa cta	
	H G D A D Y E L	
JAK2	nt	2930
	ttg ttt act cca gat tat gaa cta	
	L F T P D Y E L	
	aa	812

Nucleotide and amino acid sequence across the BCR-JAK2 fusion breakpoint.



Note that this is just the hypothetical BCR-JAK2 fusion protein. Numbers are amino acids from start of protein. The fusion protein contains the coiled-coiled domain of BCR and the kinase domain (PK1 or JH1) of JAK2.

Fusion protein

Note: The fusion protein was not detected on Western blots.

Description

The fusion protein is presumably a constitutively active kinase.

Expression localisation

Not known.

Oncogenesis

Possibly constitutive activation of the tyrosine kinase.

References

Griesinger F, Hennig H, Hillmer F, Podleschny M, Steffens R, Pies A, Wörmann B, Haase D, Bohlander SK. A BCR-JAK2 fusion gene as the result of a t(9;22)(p24;q11.2) translocation in a patient with a clinically typical chronic myeloid leukemia. *Genes Chromosomes Cancer* 2005;44:329-333.

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