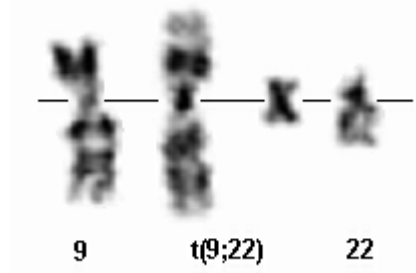


t(9;22)(p24;q11.2)

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

Gene Name [BCR1](#)

Location 22q11.2

Gene Name [JAK2](#)

Location 9p24
 Protein Janus activated kinase 2, protein tyrosine kinase

Result of the chromosomal anomaly

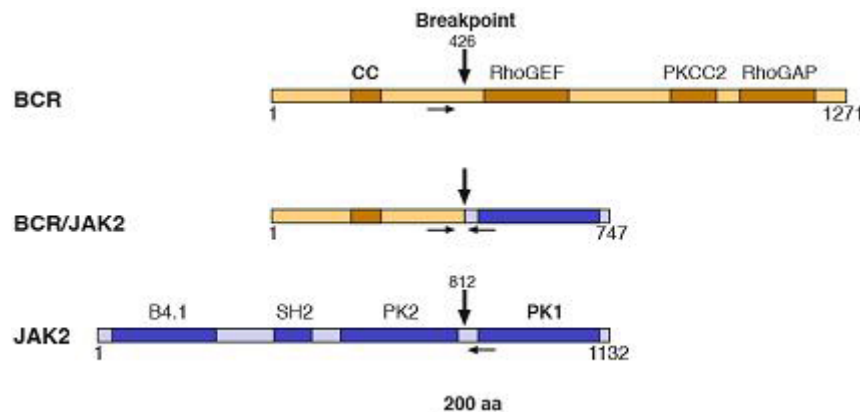
Hybrid gene Note Only the BCR-JAK2 fusion transcript was detected. The reciprocal JAK2-BCR fusion transcript could not be amplified.

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.

Detection The fusion transcript can be detected by RT-PCR using the 5' BCR sense primer: 5'-cagaactcgcaacagtccttc-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.

Fusion Protein Note The fusion protein was not detected on Western blots.



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.

Description The fusion protein is presumably a constitutively active kinase.
Oncogenesis Possibly constitutive activation of the tyrosine kinase.

External links

Other database [t\(9;22\)\(p24;q11.2\)](#) [Mitelman database \(CGAP - NCBI\)](#)
Other database [t\(9;22\)\(p24;q11.2\)](#) [CancerChromosomes \(NCBI\)](#)

To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:
you are welcome to submit a paper to our new [Case Report section](#).

Bibliography

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.

Griesinger F, Hennig H, Hillmer F, Podleschny M, Steffens R, Pies A, Wormann B, Haase D, Bohlander SK.

Genes Chromosomes Cancer 2005; 44: 329-333.

Medline [16001431](#)

Contributor(s)

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URL : <http://AtlasGeneticsOncology.org/Anomalies/t0922p24q11ID1331.html>

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