t(9;22)(p24;q11.2)  
Stefan K Bohlander  
Department of Medicine III, University Hospital Grosshadern, Marchioninistr. 15, D-81377 Munich, Germany  
or/ bGSF, Clinical Cooperative Group Leukemia, Marchioninistr. 25, D-81377 Munich, Germany  

Published in Atlas Database: November 2005  
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0922p24q11ID1331.html  
DOI: 10.4267/2042/38306  
This work is licensed under a Creative Commons Attribution-Non-commercial-No Derivative Works 2.0 France Licence.  
© 2006 Atlas of Genetics and Cytogenetics in Oncology and Haematology  

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 protocol  
The fusion transcript can be detected by RT-PCR using the 5’ BCR sense primer: 5’-cgaactcgeaagtccttc-3’ (bp 1602-1622) and the 3’ JAK2 antisense primer: 5’tcatacggcaatatc-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.
Nucleotide and amino acid sequence across the BCR-JAK2 fusion breakpoint.

**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**

*This article should be referenced as such: Bohlander SK. t(9;22)(p24;q11.2). Atlas Genet Cytogenet Oncol Haematol.2006; 10(2):123-124.*