

## t(8;9)(p22;p24)

### Clinics and Pathology

<b>Disease</b>	The PCM1-JAK2 resulting from a t(8;9)(p22;p24) fusion gene occurs in both myeloid and lymphoid malignancies: CML-like chronic phase disease with associated eosinophilia and marrow fibrosis and possible evolvement to secondary AML and B-ALL ('blast crisis'), de novo B-ALL and T-ALL/ <u>T-NHL</u> . Striking male predominance.
Phenotype / cell stem origin	<a href="#">Atypical chronic myeloid leukemia</a> ; chronic eosinophilic leukemia; pre-B-cell acute lymphoblastic leukemia; <a href="#">acute myeloid leukemia M6</a> ; T-cell acute lymphoid leukemia; myelodysplastic syndrome/myeloproliferative disease, unclassifiable; secondary acute myeloid leukemia.
Epidemiology	15 published cases (plus 3 unpublished), striking male predominance, only 2 females, median age 45.5 years (range, 12-74).
Clinics	CML-like chronic phase disease with associated eosinophilia and marrow fibrosis and possible evolvement to secondary AML and B-ALL ('blast crisis'), de novo B-ALL and T-ALL/T-NHL. Striking male predominance, clinical course highly variable.
Treatment	Allogeneic stem cell transplantation; interferon; hydroxyurea; no specific JAK2 inhibitor currently available.
Prognosis	PCM1-JAK2 positive disease is an aggressive disease compared to patients with MPD and associated V617F JAK2 mutation. Acute leukemias (de novo and secondary) seen in approximately 50% of all cases.

### Cytogenetics

Cytogenetics	t(8;9)(p22;p24).
Morphological	
Probes	First probe: 5' and 3' regions of PCM1 (RP11-49F3 and RP11-3K23). Second probe: 5' and 3' regions of JAK2 (RP11-3H3 and RP11-28A9).

### Genes involved and Proteins

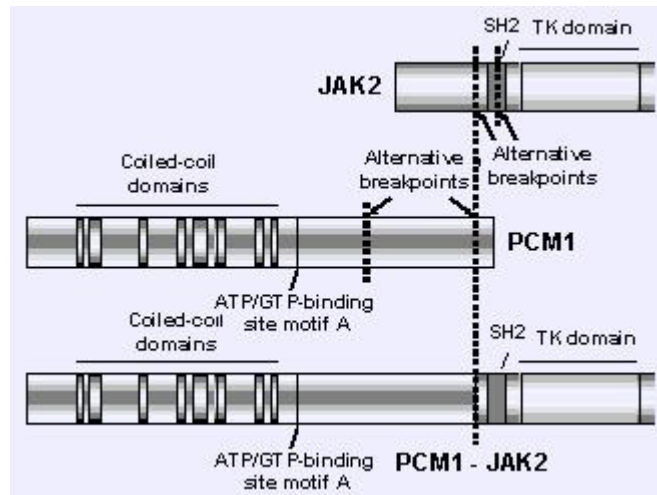
<b>Gene Name</b>	<a href="#">PCM1 (pericentriolar material 1)</a> .
Location	8p22-p21.3.
Dna / Rna	41 exons; alternate splicing.
Protein	PCM1 is involved in recruiting proteins necessary for centrosome replication and predicted to contain multiple coiled-coil motifs.
<b>Gene Name</b>	<a href="#">JAK2 (Janus kinase 2)</a> .
Location	9p24.
Dna / Rna	23 exons.
Protein	JAK2 is a tyrosine-protein kinase with transmembrane and tyrosine kinase domains.

### Result of the chromosomal anomaly

#### Hybrid gene

Description 5' PCM1 - 3' JAK2.  
 Transcript PCM1-JAK2 chimeric RNA constantly present; variable positions of the breakpoints within PCM1 and JAK2; reciprocal transcript may be present.

## Fusion Protein



Diagrammatic representation of normal JAK2, normal PCM1 and the PCM1-JAK2 fusion protein.

Description PCM1-JAK2 mRNA is predicted to encode a protein that retains several of the predicted coiled-coil domains from PCM1 and the entire tyrosine kinase domain of JAK2.

Oncogenesis As has been found for other tyrosine kinase fusion proteins, e.g. [BCR-ABL](#), it is likely that one or more of the coiled-coil motifs from PCM1 result in dimerization or oligomerization of the PCM1-JAK2 chimera, with consequent constitutive activation of the JAK2 kinase domain.

## External links

Other database [t\(8;9\)\(p22;p24\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(8;9\)\(p22;p24\)](#) [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

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## Contributor(s)

**Written** 09-2006 Andreas Reiter, Christoph Walz

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