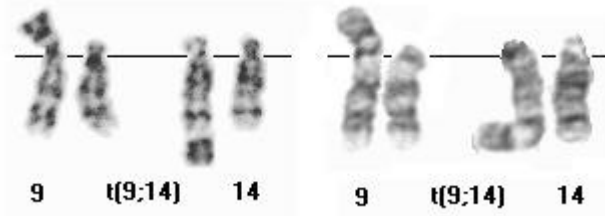


## t(9;14)(p13;q32)

### Identity



t(9;14)(p13;q32) (G- banding) left: Courtesy Bruce Poppe, Pascale De Paepe, Frank Speleman, right: Courtesy Jean-Luc Lai.

### Clinics and Pathology

**Disease** Rare recurrent chromosomal aberration, exclusively detected in B-cell lymphoproliferative disorders.

**Phenotype / cell stem origin** B lymphocyte.

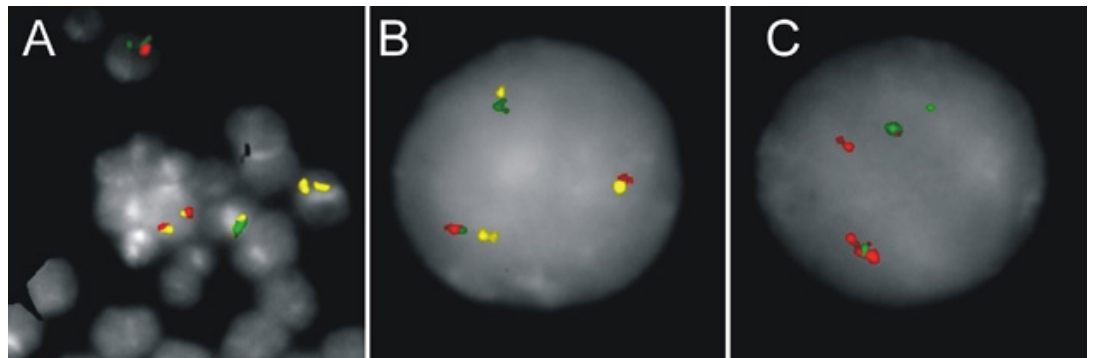
**Epidemiology** Originally reported to be associated with a low-grade mature B-cell phenotype with plasmacytoid differentiation such as lymphoplasmacytic lymphoma, [multiple myeloma](#)/ [plasma cell leukemia](#) and [chronic lymphocytic leukemia](#). However, the relatively frequent occurrence in [diffuse large B-cell lymphoma](#), with or without a preceding phase of a low-grade lymphoma, suggests that this chromosomal aberration has a much wider clinical spectrum or is associated with disease progression. In addition, the t(9;14)(p13;q32) has been described occasionally in [follicular lymphoma](#), [mantle cell lymphoma](#) and [splenic marginal zone lymphoma](#).

**Prognosis** No prognostic relevance has been attributed to the presence of the t(9;14)(p13;q32).

### Cytogenetics

**Cytogenetics** The t(9;14)(p13;q32) is readily recognisable with G- as well as R-banding.

**Morphological** The presence of complex chromosomal aberrations, however, can mask the presence of this rearrangement.



Dual and triple colour hybridisations demonstrating the presence of a t(9;14)(p13;q32) resulting in PAX5/IGH rearrangement. A,B: partial metaphase and interphase nucleus cohybridized with PAX5 locus specific probes (yellow) and dual colour interphase cytogenetics using IgH flanking probes (red) and PAX5 locus specific probes.

Probes	Probes used for detection of PAX5 rearrangements include PAX5 spanning probes (RP11-12P15, RP11-344B23 and RP11-297B17) and a probe extending approximately up to 200kb upstream of PAX5, RP11-220I1. IgH rearrangements can be demonstrated using a commercially available dual colour probe, or IgH probes, RP11-47P23 and RP5-998D24, mapping to the IgH constant and variable regions, respectively.
Additional anomalies	No recurrent additional aberrations have been described. However, the majority of t(9;14)(p13;q32) have been reported in addition to complex chromosomal aberrations.
Variants	In addition to the t(9;14)(p13;q32), other translocations presumably involving the immunoglobulin light chain genes and PAX5 have been reported, such as the t(2;9)(p12;p13) and the t(9;22)(p13;q11).

### Genes involved and Proteins

**Gene Name** IgH

Location 14q32

**Gene Name** PAX5

Location 9p13

**Dna / Rna** The PAX5 coding region extends over a genomic interval of approximately 200kb and comprises 10 exons. Two alternative transcripts have been identified, originating from alternative promoter usage, containing exon 1A or 1B. Full length mRNA is 3650bp.

**Protein** PAX5 belongs to the paired box family of transcription factors, involved in a multitude of developmental processes. PAX5 was originally identified as a B-cell specific transcription factor (hence its original name, BSAP). Recently it has been shown that PAX5 expression is continuously required in B cell lineage commitment during early B cell development.

### Result of the chromosomal anomaly

**Hybrid gene** Translocation of the entire PAX5 gene to chromosome 14. The breakpoints at 9p13 are heterogeneous and can reside up to 200kb

**Description** upstream (i.e. centromeric) of PAX5.

**Detection** The variability of the chromosomal breakpoints at 9p13 as well 14q32

precludes genomic PCR approaches for detection of IgH PAX5 juxtaposition. In addition, the expression pattern of PAX5 hampers RT-PCR methods for demonstrating elevated PAX5 expression in B-cell proliferations with suspected or proven PAX5 rearrangement. Currently, the only methods for detecting IgH PAX5 juxtaposition reliably include conventional and molecular cytogenetics.

**Fusion Protein** In analogy to other 14q32 rearrangements, no fusion gene is created by the translocation. Rather, the genomic rearrangement leads to forced PAX5 expression.  
**Description** PAX5 expression.  
**Oncogenesis** In contrast to the novel insights in the role of PAX5 in B-cell lineage commitment, little is known on the role of PAX5 in the malignant transformation of B cells. The recent demonstration of PAX5 hypermutation in diffuse large-cell lymphomas, in addition to PAX5 overexpression associated with the t(9;14), suggest that PAX5 acts as a dominant oncogene.

### External links

Other database [t\(9;14\)\(p13;q32\)](#) [Mitelman database \(CGAP - NCBI\)](#)  
Other database [t\(9;14\)\(p13;q32\)](#) [CancerChromosomes \(NCBI\)](#)  
[and ~200kb flanking regions: RP11-12P15 \(AL158148\), RP11-344B23 \(AL450267\), RP11-297B17 \(AL161781\) and RP11-22011 \(AL512604\).](#)  
[LSI® IGH Dual Color, Break Apart Rearrangement Probe, Vysis or RP11-47P23 \(AZ579057 and AQ202909\) and RP5-998D24 \(AZ579060 and AZ579059\).](#)

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