

## Gene Section

### Short Communication

# PAX5 (paired box gene 5)

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Published in Atlas Database: January 2004

Online updated version: <http://AtlasGeneticsOncology.org/Genes/PAX5ID62.html>

DOI: 10.4267/2042/38062

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## Identity

**Other names:** BSAP (B-cell lineage specific activator protein)

**HGNC (Hugo):** PAX5

**Location:** 9p13

## DNA/RNA

### Description

The PAX5 coding region extends over a genomic interval of approximately 200kb and comprises 10 exons.

### Transcription

Two alternative transcripts have been identified, originating from alternative promoter usage, containing exon 1A or 1B; full length mRNA is 3650bp; transcription is from centromere to telomere.

## Protein

### Description

391 amino acids, 42 kDa, PAX5 belongs to the paired box family of transcription factors, contains a paired box (DNA binding) domain, a truncated homeo domain homology region, and a transactivation domain.

### Expression

B lymphocytes, the developing CNS, and adult testis.

### Localisation

Nuclear.

### Function

Involved in a multitude of developmental processes, PAX5 expression is not only continuously required for B cell lineage commitment during early B cell development but also for B lineage maintenance,

involved in the regulation of the CD19 gene, a B-lymphoid-specific target gene.

## Implicated in

### *t(9;14)(p13;q23) lymphoproliferative disorders*

#### Hybrid/Mutated gene

PAX5-IGH juxtaposition.

### *dic(9;12)(p13;p13) acute lymphoblastic leukemia*

#### Hybrid/Mutated gene

PAX5-ETV6.

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*This article should be referenced as such:*

Strehl S. PAX5 (paired box gene 5). *Atlas Genet Cytogenet Oncol Haematol.* 2004; 8(2):65-66.

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