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.

References


Souabni A, Cobaleda C, Schebesta M, Busslinger M. Pax5 promotes B lymphopoiesis and blocks T cell development by repressing Notch1. Immunity. 2002 Dec;17(6):781-93


This article should be referenced as such: