

Gene Section

Mini Review

PAX5 (paired box gene 5)

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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 200 kb and comprises 10 exons.

Transcription

Two alternative transcripts have been identified, originating from alternative promotor usage, containing exon 1A or 1B; full length mRNA is 3650 bp; 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

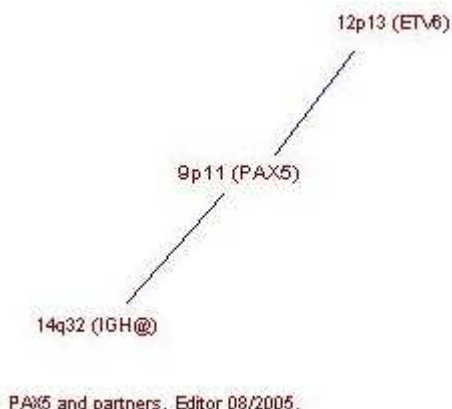
PAX5 - IGH juxtaposition.

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

Hybrid/Mutated gene

PAX5 - ETV6.

Breakpoints



References

- Barberis A, Widenhorn K, Vitelli L, Busslinger M. A novel B-cell lineage-specific transcription factor present at early but not late stages of differentiation. *Genes Dev.* 1990 May;4(5):849-59
- Offit K, Parsa NZ, Filippa D, Jhanwar SC, Chaganti RS. t(9;14)(p13;q32) denotes a subset of low-grade non-Hodgkin's lymphoma with plasmacytoid differentiation. *Blood.* 1992 Nov 15;80(10):2594-9
- Busslinger M, Klix N, Pfeffer P, Graninger PG, Kozmik Z. Deregulation of PAX-5 by translocation of the Emu enhancer of the IgH locus adjacent to two alternative PAX-5 promoters in a diffuse large-cell lymphoma. *Proc Natl Acad Sci U S A.* 1996 Jun 11;93(12):6129-34
- Iida S, Rao PH, Nallasivam P, Hibshoosh H, Butler M, Louie DC, Dyomin V, Ohno H, Chaganti RS, Dalla-Favera R. The t(9;14)(p13;q32) chromosomal translocation associated with lymphoplasmacytoid lymphoma involves the PAX-5 gene. *Blood.* 1996 Dec 1;88(11):4110-7
- Nutt SL, Urbánek P, Rolink A, Busslinger M. Essential functions of Pax5 (BSAP) in pro-B cell development: difference between fetal and adult B lymphopoiesis and reduced V-to-DJ recombination at the IgH locus. *Genes Dev.* 1997 Feb 15;11(4):476-91
- Hamada T, Yonetani N, Ueda C, Maesako Y, Akasaka H, Akasaka T, Ohno H, Kawakami K, Amakawa R, Okuma M. Expression of the PAX5/BSAP transcription factor in haematological tumour cells and further molecular characterization of the t(9;14)(p13;q32) translocation in B-cell non-Hodgkin's lymphoma. *Br J Haematol.* 1998 Aug;102(3):691-700
- Nutt SL, Heavey B, Rolink AG, Busslinger M. Commitment to the B-lymphoid lineage depends on the transcription factor Pax5. *Nature.* 1999 Oct 7;401(6753):556-62
- Ohno H, Ueda C, Akasaka T. The t(9;14)(p13;q32) translocation in B-cell non-Hodgkin's lymphoma. *Leuk Lymphoma.* 2000 Feb;36(5-6):435-45
- Cazzaniga G, Daniotti M, Tosi S, Giudici G, Aloisi A, Pogliani E, Kearney L, Biondi A. The paired box domain gene PAX5 is fused to ETV6/TEL in an acute lymphoblastic leukemia case. *Cancer Res.* 2001 Jun 15;61(12):4666-70
- Mikkola I, Heavey B, Horcher M, Busslinger M. Reversion of B cell commitment upon loss of Pax5 expression. *Science.* 2002 Jul 5;297(5578):110-3
- Schebesta M, Heavey B, Busslinger M. Transcriptional control of B-cell development. *Curr Opin Immunol.* 2002 Apr;14(2):216-23
- 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
- Hesslein DG, Pflugh DL, Chowdhury D, Bothwell AL, Sen R, Schatz DG. Pax5 is required for recombination of transcribed, acetylated, 5' IgH V gene segments. *Genes Dev.* 2003 Jan 1;17(1):37-42
- Strehl S, König M, Dworzak MN, Kalwak K, Haas OA. PAX5/ETV6 fusion defines cytogenetic entity dic(9;12)(p13;p13). *Leukemia.* 2003 Jun;17(6):1121-3
- Busslinger M. Transcriptional control of early B cell development. *Annu Rev Immunol.* 2004;22:55-79
- Fuxa M, Skok J, Souabni A, Salvaggio G, Roldan E, Busslinger M. Pax5 induces V-to-DJ rearrangements and locus contraction of the immunoglobulin heavy-chain gene. *Genes Dev.* 2004 Feb 15;18(4):411-22
- Tiacci E, Pileri S, Orleth A, Pacini R, Tabarrini A, Frenguelli F, Liso A, Diverio D, Lo-Coco F, Falini B. PAX5 expression in acute leukemias: higher B-lineage specificity than CD79a and selective association with t(8;21)-acute myelogenous leukemia. *Cancer Res.* 2004 Oct 15;64(20):7399-404
- Poppe B, De Paepe P, Michaux L, Dastugue N, Bastard C, Herens C, Moreau E, Cavazzini F, Yigit N, Van Limbergen H, De Paepe A, Praet M, De Wolf-Peeters C, Wlodarska I, Speleman F. PAX5/IGH rearrangement is a recurrent finding in a subset of aggressive B-NHL with complex chromosomal rearrangements. *Genes Chromosomes Cancer.* 2005 Oct;44(2):218-23

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