

## HLXB9 (homeo box HB9)

### Identity

Note telomeric to c7orf3 and SHH  
 Other names **HB9**  
**HOXHB9**  
**SCRA1**  
**Mnr1**  
 Hugo **HLXB9**  
 Location 7q36.3  
 Note telomeric to c7orf3 and SHH

### DNA/RNA

Description 3 exons stretched over an area of 5-6 kb.  
 Transcription In a telomere to centromere direction; 2061 bp mRNA, 1206 bp open reading frame.

### Protein

Description The homeobox gene HLXB9 encodes the nuclear protein HB9. The protein contains a polyalanine repeat region and a homeobox domain.  
 Expression Expressed in lymphoid and pancreatic tissues. Highly expressed in CD34+ bone marrow cells, down regulated upon differentiation.  
 Localisation Nuclear  
 Function Putative transcription factor.  
 Homology Related to Mnr2.

### Mutations

Note Mutations in HLXB9 cause an autosomal dominant form of sacral agenesis, known as Currarino syndrome.

### Implicated in

Entity [t\(7;12\)\(q36;p13\)](#) - associated infant acute myeloid leukemia (AML)  
 Prognosis Prognosis probably poor: median survival is 13 months.  
 Cytogenetics [t\(7;12\)\(q36;p13\)](#), but not always visible by chromosome banding; may also be misdiagnosed as [del\(12\)\(p13\)](#).

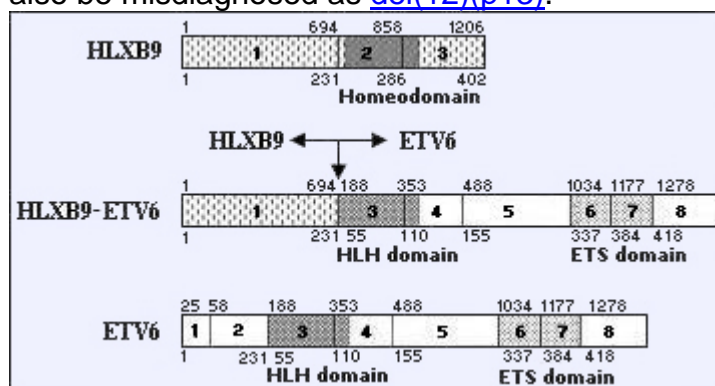


Fig. 3. Schematic representation of the HLXB9 and ETV6 proteins and the putative HLXB9-ETV6 chimeric protein resulting from the [t\(7;12\)\(q36;p13\)](#). Arrow, the observed breakpoints. nt numbers (cDNA level) are given above each protein, and

amino acid numbers are given in bold type below each protein.

Hybrid/Mutated Gene 5' HLXB9 \_ 3' [ETV6](#)  
Abnormal Protein N-term HLXB9, including its polyalanine repeat, is fused to a large C-term part of the ETV6 protein including its HLH domain and ETS domain; the homeobox domain of HLXB9 is not retained in the fusion protein; the reciprocal transcript is not expressed.

### To be noted

The t(7;12) is heterogeneous at the molecular level. The formation of a fusion gene has only been described in 2 cases and may not be the only mechanism by which HLXB9 is involved in t(7;12)-associated leukaemias. Additional 7q36 genes may also be involved.

### External links

Nomenclature	
<a href="#">Hugo</a>	<a href="#">HLXB9</a>
<a href="#">GDB</a>	<a href="#">HLXB9</a>
<a href="#">Entrez_Gene</a>	<a href="#">HLXB9 3110</a> homeobox HB9
Cards	
<a href="#">Atlas</a>	<a href="#">HLXB9ID393</a>
<a href="#">GeneCards</a>	<a href="#">HLXB9</a>
<a href="#">Ensembl</a>	<a href="#">HLXB9</a>
<a href="#">Genatlas</a>	<a href="#">HLXB9</a>
<a href="#">GeneLynx</a>	<a href="#">HLXB9</a>
<a href="#">eGenome</a>	<a href="#">HLXB9</a>
<a href="#">euGene</a>	<a href="#">3110</a>
Genomic and cartography	
<a href="#">GoldenPath</a>	<a href="#">HLXB9 - 7q36.3 chr7:156490308-156496108 - 7q36.3</a> (hg18-Mar_2006)
<a href="#">Ensembl</a>	<a href="#">HLXB9 - 7q36.3 [CytoView]</a>
<a href="#">NCBI</a>	<a href="#">Genes Cyto</a> <a href="#">Gene Seq</a> [Map View - NCBI]
<a href="#">OMIM</a>	<a href="#">Disease map [OMIM]</a>
<a href="#">HomoloGene</a>	<a href="#">HLXB9</a>
Gene and transcription	
<a href="#">Genbank</a>	<a href="#">AF107457</a> [ENTREZ]
<a href="#">Genbank</a>	<a href="#">CR623223</a> [ENTREZ]
<a href="#">Genbank</a>	<a href="#">X56537</a> [ENTREZ]
<a href="#">RefSeq</a>	<a href="#">NM_005515</a> [SRS] <a href="#">NM_005515</a> [ENTREZ]
<a href="#">AceView</a>	<a href="#">HLXB9</a> AceView - NCBI
<a href="#">TRASER</a>	<a href="#">HLXB9</a> Traser - Stanford
<a href="#">Unigene</a>	<a href="#">Hs.37035</a> [SRS] <a href="#">Hs.37035</a> [NCBI] <a href="#">HS37035</a> [spliceNest]
Protein : pattern, domain, 3D structure	
<a href="#">SwissProt</a>	<a href="#">P50219</a> [SRS] <a href="#">P50219</a> [EXPASy] <a href="#">P50219</a> [INTERPRO]
<a href="#">Prosit</a>	<a href="#">PS00027 HOMEBOX_1</a> [SRS] <a href="#">PS00027 HOMEBOX_1</a> [Expasy]
<a href="#">Prosit</a>	<a href="#">PS50071 HOMEBOX_2</a> [SRS] <a href="#">PS50071 HOMEBOX_2</a> [Expasy]
<a href="#">Interpro</a>	<a href="#">IPR001356 Homeobox</a> [SRS] <a href="#">IPR001356 Homeobox</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR012287 Homeodomain-rel</a> [SRS] <a href="#">IPR012287 Homeodomain-rel</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR009057 Homeodomain_like</a> [SRS] <a href="#">IPR009057 Homeodomain_like</a> [EBI]

[CluSTr](#) [P50219](#)  
[Pfam](#) [PF00046 Homeobox](#) [ SRS ] [PF00046 Homeobox](#) [ Sanger ] [pfam00046](#)  
 [ NCBI-CDD ]  
[Smart](#) [SM00389 HOX](#) [EMBL]  
[Prodom](#) [PD000010 Homeobox](#)[INRA-Toulouse]  
[Prodom](#) [P50219 HLXB9 HUMAN](#) [ Domain structure ] [P50219 HLXB9 HUMAN](#) [ sequences sharing at least 1 domain ]  
[Blocks](#) [P50219](#)  
[HPRD](#) [P50219](#)

### Protein Interaction databases

[DIP](#) [P50219](#)  
[IntAct](#) [P50219](#)

### Polymorphism : SNP, mutations, diseases

[OMIM](#) [142994;176450](#) [ map ]  
[GENECLINICS](#) [142994;176450](#)  
[SNP](#) [HLXB9](#) [dbSNP-NCBI]  
[SNP](#) [NM\\_005515](#) [SNP-NCI]  
[SNP](#) [HLXB9](#) [GeneSNPs - Utah] [HLXB9](#) [HGBASE - SRS]  
[HAPMAP](#) [HLXB9](#) [HAPMAP]

### General knowledge

[Family Browser](#) [HLXB9](#) [UCSC Family Browser]  
[SOURCE](#) [NM\\_005515](#)  
[SMD](#) [Hs.37035](#)  
[SAGE](#) [Hs.37035](#)  
[Amigo](#) [transcription factor activity](#)  
[Amigo](#) [RNA polymerase II transcription factor activity](#)  
[Amigo](#) [nucleus](#)  
[Amigo](#) [regulation of transcription from RNA polymerase II promoter](#)  
[Amigo](#) [humoral immune response](#)  
[Amigo](#) [morphogenesis](#)  
[Amigo](#) [sequence-specific DNA binding](#)  
[PubGene](#) [HLXB9](#)

### Other databases

#### Probes

[Probe](#) [HLXB9 Related clones \(RZPD - Berlin\)](#)

#### PubMed

[PubMed](#) [9 Pubmed reference\(s\) in LocusLink](#)

### Bibliography

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Harrison KA, Druey KM, Deguchi Y, Tuscano JM, Kehrl JH.

J Biol Chem 1994; 269(31): 19968-19975.

Medline [94327547](#)

**A homeobox gene, HLXB9, is the major locus for dominantly inherited sacral agenesis.**

Ross AJ, Ruiz-Perez V, Wang Y, Hagan DM, Scherer S, Lynch SA, Lindsay S, Custard E, Belloni E, Wilson DI, Wadey R, Goodman F, Orstavik KH, Monclair T, Robson S, Reardon W, Burn J, Scambler P, Strachan T.

Nat Genet 1998; 20(4): 358-361.

Medline [99057341](#)

**Fusion of the homeobox gene HLXB9 and the ETV6 gene in infant acute myeloid leukemias with the t(7;12)(q36;p13).**

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Cancer Res 2001; 61(14): 5374-5377

Medline [21347207](#)

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**Written**      12-2003 Anne RM von Bergh, H Berna Beverloo

**Citation**

*This paper should be referenced as such :*

**von Bergh ARM, Beverloo HB** . HLXB9 (homeo box HB9). Atlas Genet Cytogenet Oncol Haematol. December 2003 .

URL : <http://AtlasGeneticsOncology.org/Genes/HLXB9ID393.html>

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