Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Gene Section
Mini Review

MNX1 (motor neuron and pancreas homeobox 1)

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Published in Atlas Database: December 2003

Online updated version : http://AtlasGeneticsOncology.org/Genes/HLXB9ID393.html
DOI: 10.4267/2042/38041

Identity

Other names: HLXB9 (homeo box HB9); HB9; HOXHB9; SCRA1; Mnr1

HGNC (Hugo): MNX1

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

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).

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.

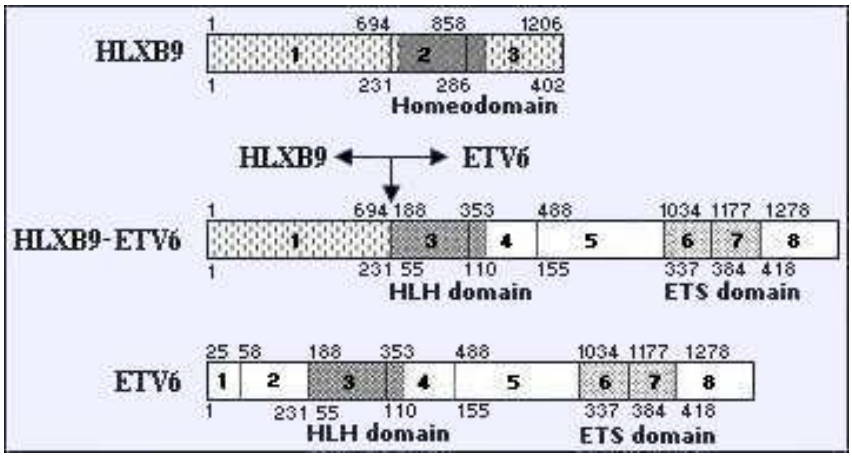

Fig. 3. Schematic representation of the HLXB9 and ETV6 proteins and the putative HLXB9-ETV6 chimeric protein resulting from the t(7;12)(q36;p13). Arrow, the observed breakpoints. nt numbers (cDNA level) are given above each protein, and amino acid numbers are given in bold type below each protein.

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.

References

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This article should be referenced as such:

von Bergh ARM, Beverloo HB. MNX1 (motor neuron and pancreas homeobox 1). Atlas Genet Cytogenet Oncol Haematol. 2004; 8(1):14-15.