

Gene Section

Mini Review

AFF3 (lymphoid nuclear protein related to AF4)

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Identity

Other names: LAF4 (lymphoid nuclear protein related to AF4); MLLT2-related protein, lymphoid nuclear protein 4

HGNC (Hugo): AFF3

Location: 2q11.2-2q12

DNA/RNA

Description

At least 17 exons.

Transcription

In a telomere to centromere direction; 3857 bp mRNA; 3684 bp open reading frame.

Protein

Description

1227 amino acids; 133734 Da.

Expression

Preferentially expressed in lymphoid tissues, highest levels being found in the thymus. Lower expression in brain and lung.

Localisation

Nuclear.

Function

Putative transcription activator that may function in lymphoid development and oncogenesis. Binds in vitro to double-stranded DNA

Homology

Belongs to the AF4/LAF4/FMR2/AF5q31 family.

Implicated in

t(2;11)(q11;q23) --> MLL-LAF4

Note

MLL fusion partner at chromosome 2 in infant ALL

Disease

The three cases reported to date were all infant-ALL with proB phenotype.

Prognosis

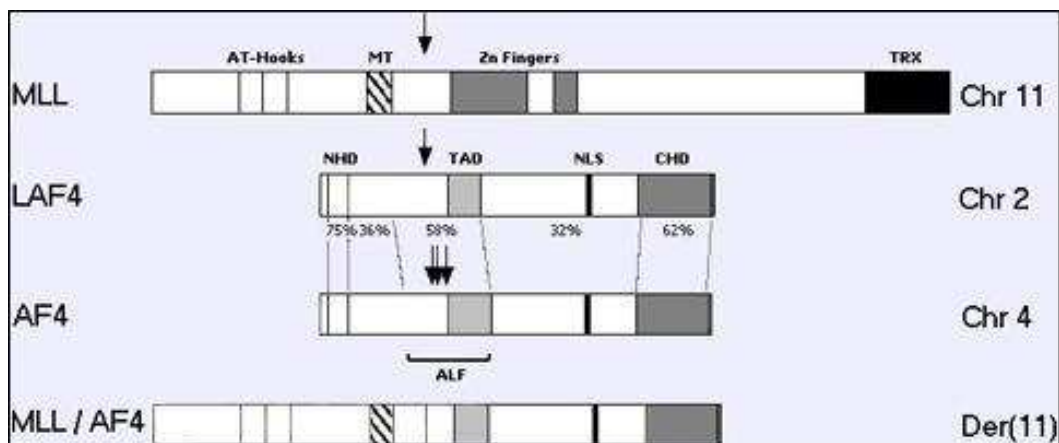
Prognosis is poor, although based on only three cases.

Cytogenetics

The three reported cases harboured different rearrangements involving chromosomes 2 and 11: *t(2;11)(q11;q23)*, *t(2;11)(p15;p14)*, and *ins(11;2)(q23;q11.2q11.2)*.

Abnormal protein

The MLL-LAF4 fusion protein includes the transactivation domain of LAF4 that is part of the AF4/LAF4/FMR2 homology domain.



Coding sequence of LAF4 compared to AF4 and site of fusion. Schematic representation of MLL, LAF4, AF4, and the putative MLL-LAF4 fusion protein. Domains in MLL are shaded: MT, DNA methyltransferase homology region; TRX, Drosophila trithorax homology. The percentage of amino acid homology between corresponding regions of LAF4 and AF4 is indicated: NHD, N-terminal homology domain; ALF, AF4/LAF4/FMR2 homology domain; TAD, transactivation domain; NLS, nuclear localization sequence; CHD, C-terminal homology domain. Arrows indicate the fusion sites with MLL. The breakpoint in LAF4 corresponds to one of the known breakpoints in AF4.

References

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