

## AF4 (ALL1 fused gene from chromosome 4) (updated: old version not available)

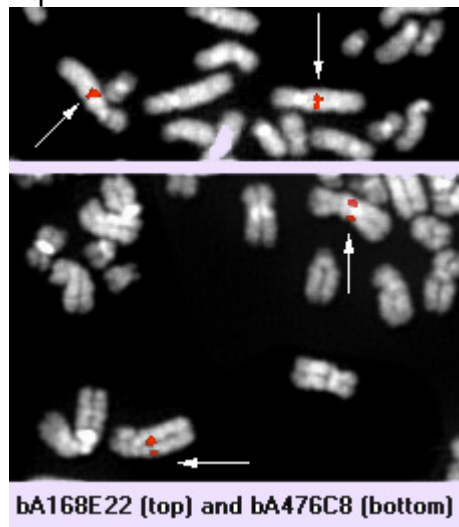
### AFF1 (AF4/FMR2 family, member 1)

#### Identity

Other names **FEL**

Hugo Location **MLLT2 (myeloid/lymphoid leukemia translocated to, 2).**

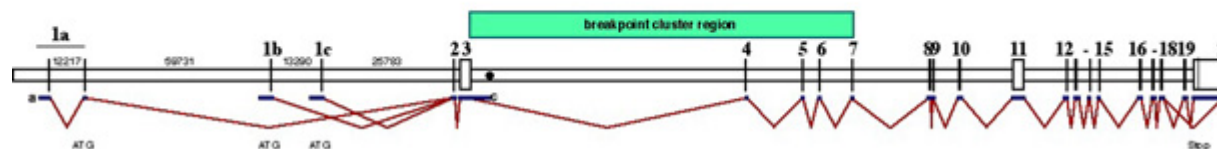
Location **MLLT2**  
4q21



[MLLT2 \(4q21\)](#) - Courtesy Mariano Rocchi, [Resources for Molecular Cytogenetics](#). Laboratories willing to validate the probes are welcome : contact [rocchi@biologia.uniba.it](mailto:rocchi@biologia.uniba.it)

#### DNA/RNA

The human AF-4 gene (4q21) consists of 23 exons



Gene structure of AF4, containing the exon/intron structure as well as the distances of all three first exons (1a is encoded by two exons; 1b and 1c) and their distances from each other (unpublished data). There is also a stop in intron 3 (as designated) and an alternative splice of exon 18 to the 3'-NTR, skipping exons 19 and 20. Therefore this protein comes in different flavors, as there are presumably three independent promoters, and one carboxy-terminal exon skipping.

Transcription alternate splicing in 5' -> 10.5 and 12 kb; coding sequences: 3.6 kb. In addition, there are three independent first exons 1a, 1b and 1c (yet unpublished).

#### Protein

Description 1210 amino acids; 140 KDa; contains many serine and proline rich sequences, a nuclear targeting sequence and a consensus sequence for ATP/GTP binding  
 Expression widely expressed  
 Localisation nuclear  
 Function transcription activator  
 Homology LAF4, AF5 and FMR-2

### Implicated in

**Entity** [t\(4;11\)\(q21;q23\)](#)/acute leukaemias. --> [MLL](#) -AF4  
**Disease** typically CD19+ B-ALL, biphenotypic AL, at times ANLL (M4/M5); may be congenital; treatment related leukaemia (secondary to epipodophyllotoxins)  
**Prognosis** median survival < 1yr  
**Cytogenetics** additional chromosome anomalies are found in 1/4 of cases of which is the i(7q)  
**Hybrid/Mutated Gene** 5' MLL-3' AF4; 12kb  
**Abnormal Protein** 240 kDa protein with about 1400 aminoacids from NH2 MLL and 850 from COOH AF4 (variable breakpoints); the reciprocal may or may not be expressed

### External links

#### Nomenclature

[Hugo](#) [MLLT2](#)  
[GDB](#) [MLLT2](#)  
[Entrez Gene](#) [MLLT2 4299](#) myeloid/lymphoid or mixed-lineage leukemia (trithorax homolog, Drosophila); translocated to, 2

#### Cards

[Atlas](#) [AF4](#)  
[GeneCards](#) [MLLT2](#)  
[Ensembl](#) [MLLT2](#)  
[CancerGene](#) [MLLT2](#)  
[Genatlas](#) [MLLT2](#)  
[GeneLynx](#) [MLLT2](#)  
[eGenome](#) [MLLT2](#)  
[euGene](#) [4299](#)

#### Genomic and cartography

[GoldenPath](#) [MLLT2 - 4q21](#) [chr4:88285319-88419369 + 4q21.3](#) (hg17-May\_2004)  
[Ensembl](#) [MLLT2 - 4q21.3 \[CytoView\]](#)  
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]  
[OMIM](#) [Disease map \[OMIM\]](#)  
[HomoloGene](#) [MLLT2](#)

#### Gene and transcription

[Genbank](#) [AJ238093](#) [SRS] [AJ238093](#) [ENTREZ]  
[Genbank](#) [X83606](#) [SRS] [X83606](#) [ENTREZ]  
[Genbank](#) [Z83689](#) [SRS] [Z83689](#) [ENTREZ]  
[Genbank](#) [L13773](#) [SRS] [L13773](#) [ENTREZ]  
[Genbank](#) [L25050](#) [SRS] [L25050](#) [ENTREZ]  
[RefSeq](#) [NM\\_005935](#) [SRS] [NM\\_005935](#) [ENTREZ]

<a href="#">RefSeq</a>	<a href="#">NT_086651</a> [SRS] <a href="#">NT_086651</a> [ENTREZ]
<a href="#">AceView</a>	<a href="#">MLLT2</a> AceView - NCBI
<a href="#">TRASER</a>	<a href="#">MLLT2</a> Traser - Stanford
<a href="#">Unigene</a>	<a href="#">Hs.480190</a> [SRS] <a href="#">Hs.480190</a> [NCBI] <a href="#">HS480190</a> [spliceNest]
<b>Protein : pattern, domain, 3D structure</b>	
<a href="#">SwissProt</a>	<a href="#">P51825</a> [SRS] <a href="#">P51825</a> [EXPASY] <a href="#">P51825</a> [INTERPRO]
<a href="#">Interpro</a>	<a href="#">IPR007797 AF-4</a> [SRS] <a href="#">IPR007797 AF-4</a> [EBI]
<a href="#">CluSTr</a>	<a href="#">P51825</a>
<a href="#">Pfam</a>	<a href="#">PF05110 AF-4</a> [SRS] <a href="#">PF05110 AF-4</a> [Sanger] <a href="#">pfam05110</a> [NCBI-CDD]
<a href="#">Blocks</a>	<a href="#">P51825</a>
<b>Polymorphism : SNP, mutations, diseases</b>	
<a href="#">OMIM</a>	<a href="#">159557</a> [ <a href="#">map</a> ]
<a href="#">GENECLINICS</a>	<a href="#">159557</a>
<a href="#">SNP</a>	<a href="#">MLLT2</a> [dbSNP-NCBI]
<a href="#">SNP</a>	<a href="#">NM_005935</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">MLLT2</a> [GeneSNPs - Utah] <a href="#">MLLT2</a> [SNP - CSHL] <a href="#">MLLT2</a> [HGBASE - SRS]
<b>General knowledge</b>	
<a href="#">Family Browser</a>	<a href="#">MLLT2</a> [UCSC Family Browser]
<a href="#">SOURCE</a>	<a href="#">NM_005935</a>
<a href="#">SMD</a>	<a href="#">Hs.480190</a>
<a href="#">SAGE</a>	<a href="#">Hs.480190</a>
<a href="#">Amigo</a>	<a href="#">component nucleus</a>
<a href="#">Amigo</a>	<a href="#">function transcription factor activity</a>
<a href="#">PubGene</a>	<a href="#">MLLT2</a>
<b>Other databases</b>	
<b>Probes</b>	
<a href="#">Probe</a>	<a href="#">Cancer Cytogenetics (Bari)</a>
<a href="#">Probe</a>	<a href="#">MLLT2 Related clones (RZPD - Berlin)</a>
<b>PubMed</b>	
<a href="#">PubMed</a>	<a href="#">8 Pubmed reference(s) in LocusLink</a>

## Bibliography

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Morrissey J, et al

Blood. 1993; 81: 1124-1131.

Medline [93184301](#)

**The t(4;11) chromosome translocation of human acute leukemias fuses the ALL-1 gene, related to Drosophila trithorax, to the AF-4 gene.**

Gu Y, et al

Cell. 1992; 71: 701-708.

Medline [93046668](#)

**Chromosome abnormalities in leukaemia: the 11q23 paradigm**

Young BD, et al

Cancer Surv. 1996; 28: 225-245. Review.

Medline [97131386](#)

**Molecular basis of 11q23 rearrangements in hematopoietic malignant proliferations.**

Bernard OA, et al

Genes Chromosomes Cancer. 1995; 13: 75-85. Review  
Medline [95352544](#)

### **11q23 rearrangements in acute leukemia.**

Rubnitz JE, et al.  
Leukemia. 1996; 10: 74-82. Review.  
Medline [96145214](#)

### **Exon/intron structure of the human AF-4 gene, a member of the AF-4/LAF-4/FMR-2 gene family coding for a nuclear protein with structural alterations in acute leukaemia.**

Nilson I, Reichel M, Ennas MG, Greim R, Knorr C, Siegler G, Greil J, Fey GH, Marschalek R.  
Br J Haematol. 1997; 98: 157-169.  
Medline [9233580](#)

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[BibliGene - INIST](#)

### **Contributor(s)**

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### **Citation**

*This paper should be referenced as such :*

**Huret JL** . AF4 (ALL1 fused gene from chromosome 4); AFF1 (AF4/FMR2 family, member 1). Atlas Genet Cytogenet Oncol Haematol. December 1997 .

URL : <http://www.infobiogen.fr/services/chromcancer/Genes/AF4.html>

**Marschalek R** . AF4 (ALL1 fused gene from chromosome 4); AFF1 (AF4/FMR2 family, member 1). Atlas Genet Cytogenet Oncol Haematol. December 2002 .

URL : <http://www.infobiogen.fr/services/chromcancer/Genes/AF4.html>

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