

Gene Section

Mini Review

AFF1 (AF4/FMR2 family, member 1)

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Huret JL. AF4 (ALL1 fused gene from chromosome 4). *Atlas Genet Cytogenet Oncol Haematol.* 1997;1(2):54-55.

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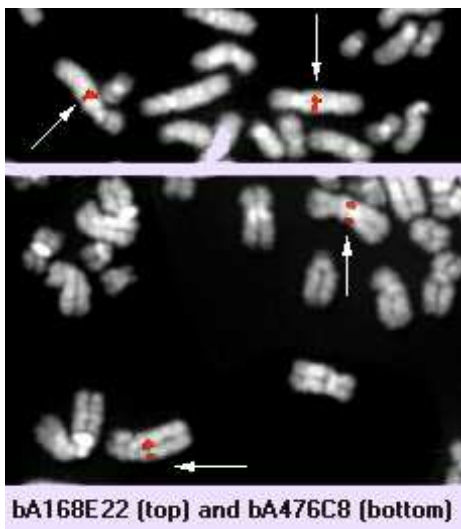
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Identity

Other names: FEL; AF4; ALL1; MLLT2 (myeloid/lymphoid leukemia translocated to, 2).

HGNC (Hugo): AFF1

Location: 4q21



MLLT2 (4q21) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

DNA/RNA

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 In addition, there are three independent first exons 1a, 1b and 1c (yet unpublished).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

t(4;11)(q21;q23)/acute leukaemias. --> MLL -AF4

Disease

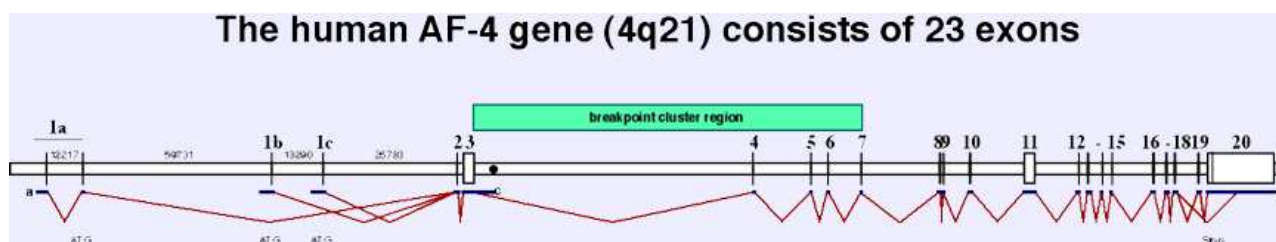
Typically CD19+ B-ALL, biphenotypic AL, at times ANLL (M4/M5); may be congenital; treatment related leukaemia (secondary to epipodophyllotoxins).

Prognosis

Median survival <1yr.

Cytogenetics

Additional chromosome anomalies are found in 1/4 of cases of which is the i(7q).



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as the distances of all three first exons (1a is encoded by two exons; 1b and 1c) and their distances from each other (unpublished data).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 exon 19 and 20. Therefore this protein comes in different flavors, as there are presumably three independent promoter, and one carboxy-terminal exon skipping.

Hybrid/Mutated gene

5' MLL - 3' AF4; 12 kb.

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.

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