AF5Q31 (ALL1 fused gene from chromosome 5q31)

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Identity

Other names: AF5q31 (ALL1 fused gene from chromosome 5q31)
HGNC (Hugo): AFF4
Location: 5q31

DNA/RNA

Description
At least 16 exons.

Transcription
In a telomere to centromere direction; 4235 bp mRNA; open reading frame: 3491 bp.

Protein

Description
1163 amino acids; 127 kDa.

Expression
Mostly in fetal tissues (heart, lung, brain, liver); at a low level in adult tissues; therefore, AF5q31 may play a critical role in the fetal development.

Homology

With AF4-related proteins: AF4, the gene involved int(4;11)(q21;q23), LAF4, FMR2.

Implicated in

ins(5;11)(q31;q13q23) acute lymphoblastic leukemia (ALL) --> MLL-AF5q31

Note
Poorly defined: only 2 cases to date, infants with CD19+ ALL; complete remission, relapse and death.

Hybrid/Mutated gene
AF5Q31 and MLL are transcribed in opposite directions, and inverted insertion is required.

References

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