

Gene Section

Mini Review

DAB2IP (DAB2 interacting protein)

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Identity

Other names: AF9q34; DIP1/2,; KIAA1743

HGNC (Hugo): DAB2IP

Location: 9q34



Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

Note: Centromeric of ABL.

DNA/RNA

Description

14 exons stretched over an area of about 84 kb. Exon 1, a noncoding exon, has (at least) three variants.

Transcription

In a centromere to telomere direction 5192 bp mRNA; 2903 bp open reading frame.

Protein

Description

The AF9q34 protein contains a GAP related domain (GRD), an 'FLR'-motif, a Pleckstrin homo-logy (PH) domain and a calcium/phospho-lipid-binding C2/CALB domain; 967 amino acids.

Expression

Expression in all human tissues with a relatively low level of expression in testis, placenta, spleen and peripheral blood leukocytes.

Function

RasGAPs negatively regulate the activity of Ras proteins that modulate diverse cellular processes by cycling between the inactive GDP-bound and active GTP-bound state. DAB2 interacting protein.

Homology

Homologous to human nGAP (or RAS protein activator like 2 (RASAL2)).

Implicated in

t(9;11)(q34;q23) AML --> MLL-AF9q34

Note

Another *t(9;11)(q34;q23)* AML has been reported with involvement of MLL-FBP17.

Disease

Only one case to date, a 62 yr old male patient with Acute Myeloid Leukemia (M5).

Cytogenetics

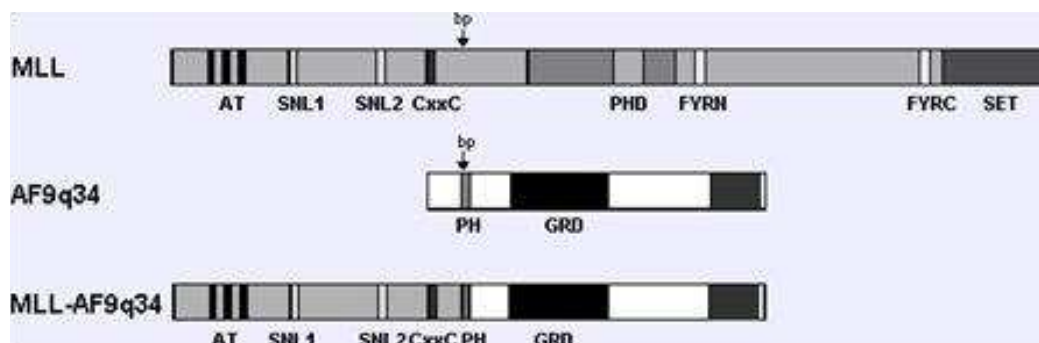
The case was 46, XY, *t(9;11)(q34;q23)* and in 40% of the metaphases additional chromosomes 8 and 13 were observed.

Hybrid/Mutated gene

5' MLL fused at exon 9 with the 3' end of AF9q34.

Abnormal protein

The MLL-AF9q34 protein includes the entire GAP related domain (GRD) and the C2/CALB domain, but the PH domain is disrupted by the breakpoint in AF9q34.



Schematic representation of MLL, AF9q34, and the putative MLL-AF9q34 fusion protein.

Domains in MLL and AF9q34 are shaded: AT, AT-hook DNA binding motifs; SNL1 and SNL2, speckled nuclear localisation signals 1 and 2; CxxC, cysteine rich motif homologous to DNA methyltransferase (MT); PHD, plant homeodomain fingers; FYRN, FYRN domain; FYRC, FYRC domain; SET, SET domain; PH, Pleckstrin Homology Domain; GRD, GAP Related Domain. Arrows indicate the breakpoint (bp) sites in MLL and AF9q34.

References

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