

## DAB2IP (DAB2 interacting protein)

### Identity

Note centromeric of [ABL](#)

Other names **AF9q34**

**DIP1/2,**

**KIAA1743**

Hugo **[DAB2IP](#)**

Location 9q34

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 homology (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)).

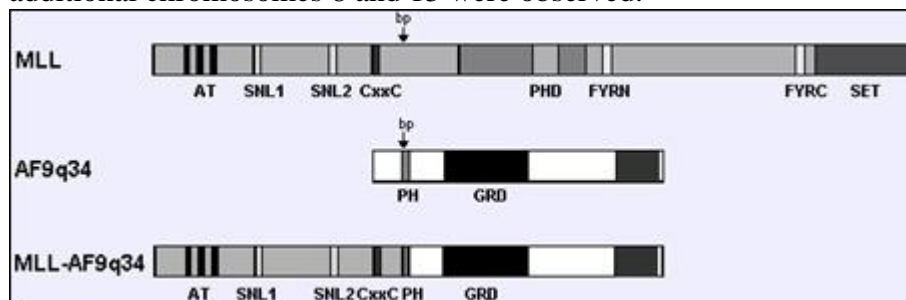
### Implicated in

Entity [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.



### 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.

Hybrid/Mutated Gene

5' MLL fused at exon 9 with the 3' end of AFq34.

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.

### External links

#### Nomenclature

[Hugo](#)

[DAB2IP](#)

[GDB](#)

[DAB2IP](#)

[Entrez\\_Gene](#)

[DAB2IP\\_153090](#) DAB2 interacting protein

#### Cards

[Atlas](#)

[AF9q34ID316](#)

[GeneCards](#)

[DAB2IP](#)

[Ensembl](#)

[DAB2IP](#)

[GenAtlas](#)

[DAB2IP](#)

[GeneLynx](#)

[DAB2IP](#)

[eGenome](#)

[DAB2IP](#)

[euGene](#)

[153090](#)

#### Genomic and cartography

[GoldenPath](#)

[DAB2IP - 9q34](#) [chr9:123501470-123587628 + 9q33.2](#) (hg18-Mar\_2006)

[Ensembl](#)

[DAB2IP - 9q33.2 \[CytoView\]](#)

[NCBI](#)

[Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]

[OMIM](#)

[Disease map \[OMIM\]](#)

[HomoloGene](#)

[DAB2IP](#)

#### Gene and transcription

[Genbank](#)

[AB051530](#) [ENTREZ]

[Genbank](#)

[AF367051](#) [ENTREZ]

[Genbank](#)

[AK054851](#) [ENTREZ]

[Genbank](#)

[AK096391](#) [ENTREZ]

[Genbank](#)

[AK124610](#) [ENTREZ]

[RefSeq](#)

[NM\\_032552](#) [SRS] [NM\\_032552](#) [ENTREZ]

[RefSeq](#)

[NM\\_138709](#) [SRS] [NM\\_138709](#) [ENTREZ]

[AceView](#)

[DAB2IP](#) AceView - NCBI

[TRASER](#)

[DAB2IP](#) Traser - Stanford

[Unigene](#)

[Hs.522378](#) [SRS] [Hs.522378](#) [NCBI] [HS522378](#) [spliceNest]

#### Protein : pattern, domain, 3D structure

[SwissProt](#)

[Q5T4Q1](#) [SRS] [Q5T4Q1](#) [EXPASY] [Q5T4Q1](#) [INTERPRO]

[Prosite](#)

[PS50003 PH DOMAIN](#) [SRS] [PS50003 PH DOMAIN](#) [Expasy]

[Interpro](#)

[IPR000008 C2\\_Ca-dep](#) [SRS] [IPR000008 C2\\_Ca-dep](#) [EBI]

[Interpro](#)

[IPR008973 C2\\_CaLB](#) [SRS] [IPR008973 C2\\_CaLB](#) [EBI]

[Interpro](#)

[IPR001849 PH](#) [SRS] [IPR001849 PH](#) [EBI]

[Interpro](#) [IPR011993 PH\\_type](#) [ SRS ] [IPR011993 PH\\_type](#) [ EBI ]  
[CluSTr](#) [Q5T4Q1](#)  
[Pfam](#) [PF00168 C2](#) [ SRS ] [PF00168 C2](#) [ Sanger ] [pfam00168](#) [ NCBI-CDD ]  
[Pfam](#) [PF00169 PH](#) [ SRS ] [PF00169 PH](#) [ Sanger ] [pfam00169](#) [ NCBI-CDD ]  
[Smart](#) [SM00239 C2](#) [EMBL]  
[Smart](#) [SM00233 PH](#) [EMBL]  
[Blocks](#) [Q5T4Q1](#)  
[HPRD](#) [Q5T4Q1](#)

**Protein Interaction databases**

[DIP](#) [Q5T4Q1](#)  
[IntAct](#) [Q5T4Q1](#)

**Polymorphism : SNP, mutations, diseases**

[OMIM](#) [609205](#) [ map ]  
[GENECLINICS](#) [609205](#)  
[SNP](#) [DAB2IP](#) [dbSNP-NCBI]  
[SNP](#) [NM\\_032552](#) [SNP-NCI]  
[SNP](#) [NM\\_138709](#) [SNP-NCI]  
[SNP](#) [DAB2IP](#) [GeneSNPs - Utah] [DAB2IP](#) [HGBASE - SRS]  
[HAPMAP](#) [DAB2IP](#) [HAPMAP]

**General knowledge**

[Family Browser](#) [DAB2IP](#) [UCSC Family Browser]  
[SOURCE](#) [NM\\_032552](#)  
[SOURCE](#) [NM\\_138709](#)  
[SMD](#) [Hs.522378](#)  
[SAGE](#) [Hs.522378](#)  
[Amigo](#) [GTPase activator activity](#)  
[Amigo](#) [intracellular](#)  
[Amigo](#) [regulation of small GTPase mediated signal transduction](#)  
[PubGene](#) [DAB2IP](#)

**Other databases**

**Probes**

[Probe](#) [clones RP6-10F14 and RP6-105L9](#)  
[Probe](#) [DAB2IP Related clones \(RZPD - Berlin\)](#)

**PubMed**

[PubMed](#) [15 Pubmed reference\(s\) in LocusLink](#)

**Bibliography**

**Cloning of unknown MLL fusion transcripts identifies two novel MLL fusion partners.**  
 von Bergh A, Beverloo B, Slater R, Groot A, Rombout P, Kluin P, Schuurin E.  
 Blood 2000; 96 Suppl 1: Abst 2984.

**Differential regulation of the human gene DAB2IP in normal and malignant prostatic epithelia: cloning and characterization.**

Chen H, Pong RC, Wang Z, Hsieh JT.  
 Genomics 2002; 79(4): 573-581.

Medline [21945266](#)

[REVIEW articles](#) *automatic search in PubMed*

[Last year](#) *automatic search in PubMed*

publications

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