

## GRAF (GTPase activating protein for Rho associated with FAK)

### Identity

Other names **KIAA0621**

**OPHN1L**

Location 5q31  
just centromeric of GRL

### DNA/RNA

Description at least 24 exons

Transcription two isoforms of 2277 bp (leukocytes) and 2442 bp (brain); transcripts of 4,4 and 9,5 kb

### Protein

▼ Tyrosine kinase phosphorylation site (Tyr<sup>141</sup>+Tyr<sup>411</sup>)

▼ N-Myristoylation site (Gly<sup>308</sup>+Gly<sup>742</sup>)

▼ Phosphorylation (Ser<sup>685</sup>) by MAP Kinase



■ Nuclear Targeting sequence (115-131)

■ PH domain (266-369)

■ GAP domain (389-561)

■ SH3 domain (706-757)

■ Immunoglobulin MHC Protein recognition sequence (710-716)

Description isoform A: 759 amino acids, 86 kDa; isoform B: 814 amino acids. 92 kDa

Expression highly expressed in epithelial tissues i.e. pancreas islet beta-cells, testicles, prostate, mammary gland, GI glands, squamous layer of skin epithelium; highly expressed in nervous tissues including enteric ganglia; expressed in cardiomyocytes, erythropoiesis cells and liver

Localisation mainly cytoplasmatic

Function interacts with FAK and [RhoA](#) both in vivo and in vitro; acts as GTPase activating protein (GAP) for the active GTP-bound RhoA.; negative regulator of RhoA

Homology Oligophrenin-1, Beta-chimerin, [BCR](#)

### Mutations

Germinal not known

Somatic deletion of four bases (251-254, A in ATG=nt1)

Insert 1158  
 GRAF-base 1144,  
 5' 1 TA GAG ACA GGA TTT CAT CAT GTT GGC CAG GTT GGT TTT GAA  
 42 TTC CTG ACC TCA AGT GAT CCA CCT GCC TCG GCC TCC CAA AGT  
 84 GGT GGG ATT TTG G 3'  
 .....GRAF-base 1145  
 Insert 1299  
 GRAF-base 1285,  
 5' 1 TC ATC GTT GTC ATA TAA ATC GGC GAG GTA ATA TTC CAT CAG  
 42 GTA GAC ATA CG 3'  
 ...GRAF-base 1286. Predicted STOP codon underlined.  
 Insert 2002  
 GRAF-base 1988  
 5' 1 G GTT CAT GCG AGT TCA GCA AGC AGT TAC CAT GTC TAC GGC  
 41 ATG CCA GGA TAC TGT TGG GAA GGT AGT ATT CCG T 3'  
 ...GRAF-base 1989

### Implicated in

**Entity** [t\(5;11\)\(q31;q23\)](#) / acute non lymphocytic leukemia --> [MLL](#) - GRAF  
**Disease** acute non lymphocytic leukemia and myelodysplastic syndrome with del(5q)  
**Prognosis** unknown; only a few cases unknown  
**Cytogenetics** [del\(5q\)](#)  
**Hybrid/Mutated Gene** 5' MLL 3' GRAF  
**Abnormal Protein** MLL-GRAF  
**Oncogenesis** basically unknown; a bi-allelic loss of GRAF has been documented in three cases of ANLL

### External links

**Nomenclature**  
[GDB](#) [ARHGAP26](#)  
[Entrez Gene](#) [ARHGAP26](#) [23092](#) Rho GTPase activating protein 26  
**Cards**  
[Atlas](#) [GRAFID291](#)  
[GeneCards](#) [ARHGAP26](#)  
[Ensembl](#) [ARHGAP26](#)  
[CancerGene](#) [GRAF](#)  
[Genatlas](#) [ARHGAP26](#)  
[GeneLynx](#) [ARHGAP26](#)  
[eGenome](#) [ARHGAP26](#)  
[euGene](#) [23092](#)  
**Genomic and cartography**  
[GoldenPath](#) [ARHGAP26](#) - [5q31](#) [chr5:142130476-142586243](#) + [5q31.3](#) (hg17-May\_2004)  
[Ensembl](#) [ARHGAP26](#) - [5q31.3](#) [[CytoView](#)]  
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [[Map View](#) - NCBI]  
[OMIM](#) [Disease map](#) [[OMIM](#)]  
[HomoloGene](#) [ARHGAP26](#)  
**Gene and transcription**  
[Genbank](#) [AF196313](#) [[SRS](#)] [AF196313](#) [[ENTREZ](#)]

[Genbank](#) [AF508552](#) [SRS] [AF508552](#) [ENTREZ]  
[Genbank](#) [Y10388](#) [SRS] [Y10388](#) [ENTREZ]  
[Genbank](#) [AB014521](#) [SRS] [AB014521](#) [ENTREZ]  
[Genbank](#) [AK092488](#) [SRS] [AK092488](#) [ENTREZ]  
[RefSeq](#) [NM\\_015071](#) [SRS] [NM\\_015071](#) [ENTREZ]  
[RefSeq](#) [NT\\_086681](#) [SRS] [NT\\_086681](#) [ENTREZ]  
[AceView](#) [ARHGAP26](#) AceView - NCBI  
[TRASER](#) [ARHGAP26](#) Traser - Stanford  
[Unigene](#) [Hs.293593](#) [SRS] [Hs.293593](#) [NCBI] [HS293593](#) [spliceNest]

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

[SwissProt](#) [Q9UNA1](#) [SRS] [Q9UNA1](#) [EXPASY] [Q9UNA1](#) [INTERPRO]  
[CluSTR](#) [Q9UNA1](#)  
[Blocks](#) [Q9UNA1](#)

### Polymorphism : SNP, mutations, diseases

[OMIM](#) [605370](#) [[map](#)]  
[GENECLINICS](#) [605370](#)  
[SNP](#) [ARHGAP26](#) [dbSNP-NCBI]  
[SNP](#) [NM\\_015071](#) [SNP-NCI]  
[SNP](#) [ARHGAP26](#) [GeneSNPs - Utah] [ARHGAP26](#) [SNP - CSHL] [ARHGAP26](#) [HGBASE - SRS]

### General knowledge

[Family Browser](#) [ARHGAP26](#) [UCSC Family Browser]  
[SOURCE](#) [NM\\_015071](#)  
[SMD](#) [Hs.293593](#)  
[SAGE](#) [Hs.293593](#)  
[Amigo](#) [function|Rho GTPase activator activity](#)  
[Amigo](#) [process|actin cytoskeleton organization and biogenesis](#)  
[Amigo](#) [component|cellular component unknown](#)  
[Amigo](#) [process|neurogenesis](#)  
[PubGene](#) [ARHGAP26](#)

### Other databases

Other database [HUGE: A Database of Human Unidentified Gene-Encoded Large Proteins](#)

### Probes

### PubMed

[PubMed](#) [7 Pubmed reference\(s\) in LocusLink](#)

### Bibliography

**The human GRAF gene is fused to MLL in a unique t(5;11)(q31;q23) and both alleles are disrupted in three cases of myelodysplastic syndrome/acute myeloid leukemia with a deletion 5q.**

Borkhardt A, Bojesen S, Haas OA, Fuchs U, Bartelheimer D, Loncarevic IF, Bohle RM, Harbott J, Repp R, Jaeger U, Viehmann S, Henn T, Korth P, Scharr D, Lampert F.

Proc Natl Acad Sci U S A. 2000; 97: 9168-9173.

Medline [20381355](#)

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## BiblioGene - INIST

### **Contributor(s)**

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### **Citation**

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URL : <http://www.infobiogen.fr/services/chromcancer/Genes/GRAFID291.html>

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