

## Gene Section

### Mini Review

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

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## Identity

**Other names:** GRAF (GTPase activating protein for Rho associated with FAK); KIAA0621; OPHN1L

**HGNC (Hugo):** ARHGAP26

**Location:** 5q31

**Local order:** 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

### Description

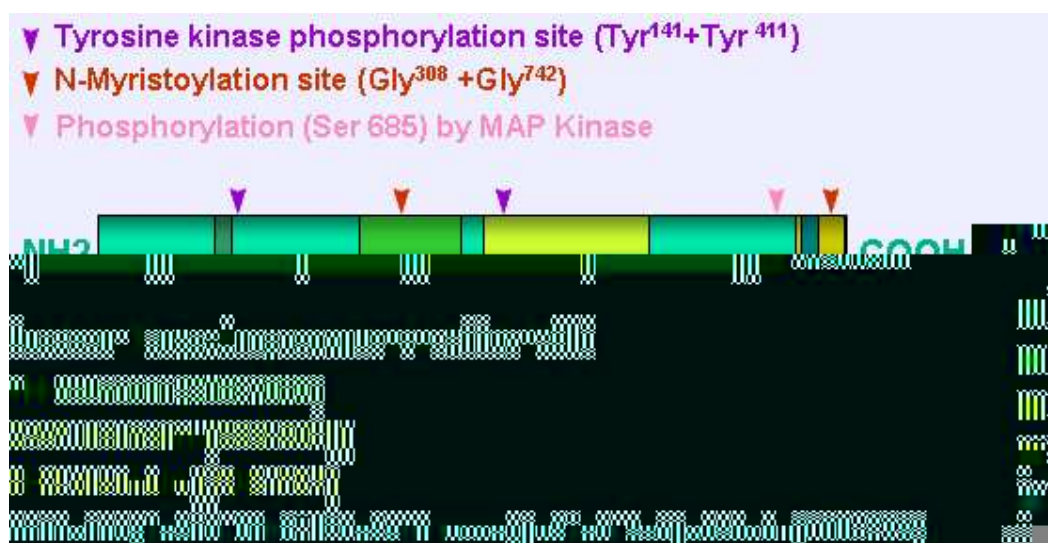
Isoform A: 759 amino acids, 86 kDa; isoform B: 814 amino acids, 92 kDa.

### 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**

***t(5;11)(q31;q23) / acute non lymphocytic leukemia --> MLL - GRAF***

**Prognosis**

Unknown; only a few cases.

**Hybrid/Mutated gene**

5' MLL 3' GRAF

**Abnormal protein**

MLL-GRAF

***Acute non lymphocytic leukemia and myelodysplastic syndrome with del(5q)***

**Prognosis**

Unknown.

**Cytogenetics**

del(5q).

**Oncogenesis**

Basically unknown; a bi-allelic loss of GRAF has been documented in three cases of ANLL.

**References**

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. 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. Proc Natl Acad Sci U S A. 2000 Aug 1;97(16):9168-73

*This article should be referenced as such:*

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