

## ARHGEF12 (Rho guanine nucleotide exchange factor (GEF) 12)

### Identity

Other names	<b>LARG (Leukemia Associated Rho Guanine nucleotide exchange factor)</b> <b>KIAA0382</b>
Hugo	<a href="#">ARHGEF12</a>
Location	11q23 telomeric to MLL

### DNA/RNA

Transcription 9501 bp mRNA; 4634 bp open reading frame

### Protein

Description	the gene encodes a guanine nucleotide exchange factor; 1544 amino acids; NH2- PDZ domain, Lsc homology (LH) domain, bipartite nuclear localization signal, Dbl homology (DH) domain, and a pleckstrin homology (PH) domain -COOH
Expression	wide (leukocytes, spleen, prostate, testis, ovary, small intestine, colon, thymus)
Function	PDZ domains are involved in protein-protein interactions in transmembrane signaling pathways; LH domains bind G proteins; nuclear localization signals are implicated in translocation of proteins from the cytoplasm to the nucleus; DH domains are responsible for the nucleotide exchange activity of guanine nucleotide exchange (GEF) toward Rho GTPases, PH domains function in membrane localization; Rho GEF are regulators of GTPases, and have oncogenic properties
Homology	member of the family of Rho GEF factors, like dbi, vav, tiam, and <a href="#">BCR</a>

### Implicated in

Entity	acute non lymphocytic leukemia (ANLL) with apparently normal chromosomes 11 --> <a href="#">MLL</a> - LARG
Note	poorly defined: only 1 case to date, a 38 yr old male patient with occupational exposure; M4-ANLL; CR; death unrelated to the leukemia
Cytogenetics	the case was 51,XY,+8,+19,+3mar; the interstitial deletion at 11q23 resulting in MLL-LARG fusion cannot be seen at the chromosome level

Hybrid/Mutated Gene      5' MLL fused at exon 6 with the 3' end of almost the entire LARG;  
 LARG is oriented in a 5' to 3' direction, like MLL

Abnormal Protein      excludes the NH2- PDZ domain of LARG, and includes most of  
 LARG, from the Lsc homology (LH) domain to COOH

## External links

### Nomenclature

[Hugo](#)      [ARHGEF12](#)

[GDB](#)      [ARHGEF12](#)

[Entrez Gene](#)      [ARHGEF12](#) [23365](#) Rho guanine nucleotide exchange factor (GEF)  
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### Cards

[Atlas](#)      [LARGID243](#)

[GeneCards](#)      [ARHGEF12](#)

[Ensembl](#)      [ARHGEF12](#)

[Genatlas](#)      [ARHGEF12](#)

[GeneLynx](#)      [ARHGEF12](#)

[eGenome](#)      [ARHGEF12](#)

[euGene](#)      [23365](#)

### Genomic and cartography

[GoldenPath](#)      [ARHGEF12](#) - [11q23](#) [chr11:119713156-119865855](#) + [11q23.3](#)  
 (hg18-Mar\_2006)

[Ensembl](#)      [ARHGEF12](#) - [11q23.3](#) [CytoView]

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

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

[HomoloGene](#)      [ARHGEF12](#)

### Gene and transcription

[Genbank](#)      [AB002380](#) [ENTREZ]

[Genbank](#)      [AF180681](#) [ENTREZ]

[Genbank](#)      [AL137456](#) [ENTREZ]

[Genbank](#)      [BC063117](#) [ENTREZ]

[Genbank](#)      [BX647785](#) [ENTREZ]

[RefSeq](#)      [NM\\_015313](#) [SRS] [NM\\_015313](#) [ENTREZ]

[AceView](#)      [ARHGEF12](#) AceView - NCBI

[TRASER](#)      [ARHGEF12](#) Traser - Stanford

[Unigene](#)      [Hs.24598](#) [SRS] [Hs.24598](#) [NCBI] [HS24598](#) [spliceNest]

## Protein : pattern, domain, 3D structure

<a href="#">SwissProt</a>	<a href="#">Q9NZN5</a> [SRS] <a href="#">Q9NZN5</a> [EXPASY] <a href="#">Q9NZN5</a> [INTERPRO]
<a href="#">Prosite</a>	<a href="#">PS00741 DH_1</a> [SRS] <a href="#">PS00741 DH_1</a> [Expasy]
<a href="#">Prosite</a>	<a href="#">PS50010 DH_2</a> [SRS] <a href="#">PS50010 DH_2</a> [Expasy]
<a href="#">Prosite</a>	<a href="#">PS50106 PDZ</a> [SRS] <a href="#">PS50106 PDZ</a> [Expasy]
<a href="#">Prosite</a>	<a href="#">PS50003 PH DOMAIN</a> [SRS] <a href="#">PS50003 PH DOMAIN</a> [Expasy]
<a href="#">Interpro</a>	<a href="#">IPR001331 GDS_CDC24</a> [SRS] <a href="#">IPR001331 GDS_CDC24</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR001478 PDZ</a> [SRS] <a href="#">IPR001478 PDZ</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR001849 PH</a> [SRS] <a href="#">IPR001849 PH</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR011993 PH_type</a> [SRS] <a href="#">IPR011993 PH_type</a> [EBI]
<a href="#">Interpro</a>	<a href="#">IPR000219 RhoGEF</a> [SRS] <a href="#">IPR000219 RhoGEF</a> [EBI]
<a href="#">CluSTr</a>	<a href="#">Q9NZN5</a>
<a href="#">Pfam</a>	<a href="#">PF00595 PDZ</a> [SRS] <a href="#">PF00595 PDZ</a> [Sanger] <a href="#">pfam00595</a> [NCBI-CDD]
<a href="#">Pfam</a>	<a href="#">PF00621 RhoGEF</a> [SRS] <a href="#">PF00621 RhoGEF</a> [Sanger] <a href="#">pfam00621</a> [NCBI-CDD]
<a href="#">Smart</a>	<a href="#">SM00228 PDZ</a> [EMBL]
<a href="#">Smart</a>	<a href="#">SM00233 PH</a> [EMBL]
<a href="#">Smart</a>	<a href="#">SM00325 RhoGEF</a> [EMBL]
<a href="#">Blocks</a>	<a href="#">Q9NZN5</a>
<a href="#">PDB</a>	<a href="#">1TXD</a> [SRS] <a href="#">1TXD</a> [PdbSum], <a href="#">1TXD</a> [IMB]
<a href="#">PDB</a>	<a href="#">1X86</a> [SRS] <a href="#">1X86</a> [PdbSum], <a href="#">1X86</a> [IMB]

## Protein Interaction databases

<a href="#">DIP</a>	<a href="#">Q9NZN5</a>
<a href="#">IntAct</a>	<a href="#">Q9NZN5</a>

## Polymorphism : SNP, mutations, diseases

<a href="#">OMIM</a>	<a href="#">601626:604763</a> [map]
<a href="#">GENECLINICS</a>	<a href="#">601626:604763</a>
<a href="#">SNP</a>	<a href="#">ARHGEF12</a> [dbSNP-NCBI]
<a href="#">SNP</a>	<a href="#">NM_015313</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">ARHGEF12</a> [GeneSNPs - Utah] <a href="#">ARHGEF12</a> [HGBASE - SRS] <a href="#">ARHGEF12</a> [SNP - HAPMAP]

## General knowledge

<a href="#">Family Browser</a>	<a href="#">ARHGEF12</a> [UCSC Family Browser]
<a href="#">SOURCE</a>	<a href="#">NM_015313</a>
<a href="#">SMD</a>	<a href="#">Hs.24598</a>
<a href="#">SAGE</a>	<a href="#">Hs.24598</a>
<a href="#">Amigo</a>	<a href="#">Rho guanyl-nucleotide exchange factor activity</a>
<a href="#">Amigo</a>	<a href="#">GTPase activator activity</a>

[Amigo](#) [protein binding](#)  
[Amigo](#) [intracellular](#)  
[Amigo](#) [regulation of Rho protein signal transduction](#)  
[PubGene](#) [ARHGEF12](#)

#### Other databases

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

#### Probes

[Probe](#) [ARHGEF12 Related clones \(RZPD - Berlin\)](#)

#### PubMed

[PubMed](#) [14 Pubmed reference\(s\) in LocusLink](#)

### Bibliography

**Identification of a gene at 11q23 encoding a guanine nucleotide exchange factor: Evidence for its fusion with MLL in acute myeloid leukemia.**

Kourlas PJ, Strout MP, Becknell B, Veronese ML, Croce CM, Theil KS, Krahe R, Ruutu T, Knuutila S, Bloomfield CD, Caligiuri MA.

Proc Natl Acad Sci USA 2000; 97: 2145-2150.

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

[BiblioGene - INIST](#)

### Contributor(s)

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URL : <http://AtlasGeneticsOncology.org/Genes/LARGID243.html>

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