

## Gene Section

### Mini Review

# ARHGAP20 (Rho GTPase activating protein 20)

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Published in Atlas Database: August 2006

Online updated version: <http://AtlasGeneticsOncology.org/Genes/ARHGAP20ID42979ch11q23.html>  
DOI: 10.4267/2042/38373

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## Identity

**Hugo:** ARHGAP20

**Other names:** KIAA1391; RARHOGAP

**Location:** 11q23.1

**Local order:** Telomeric to ATM.

## DNA/RNA

### Description

19 exons spanning 136.1 kb genomic DNA.

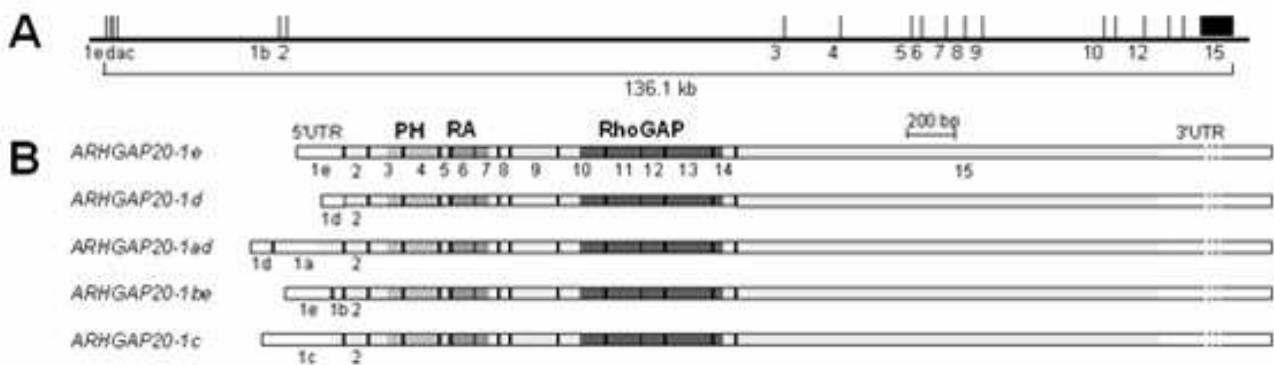
### Transcription

5.9-6.2 kb mRNA, coding sequence: 3.5-3.6 kb

Alternative splicing of the first 5 exons results in the expression of 5 transcript variants (ARHGAP20-1e, ARHGAP20-1d, ARHGAP20-1ad, ARHGAP20-1be, ARHGAP20-1c).

### Pseudogene

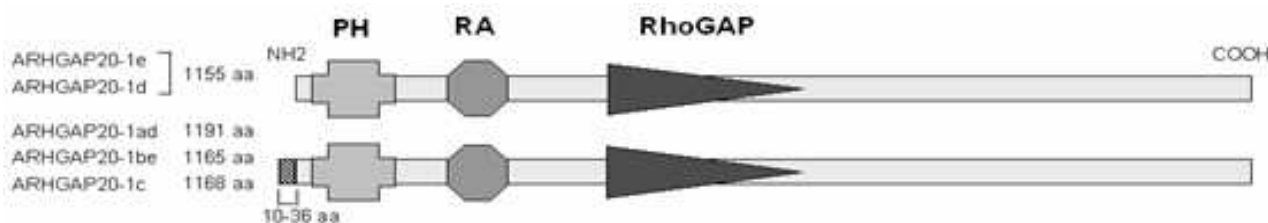
None.



Genomic organization (A) and transcript variants (B) of ARHGAP20.

(A) Gene structure (drawn to scale): black boxes represent exons.

(B) Transcripts (drawn to scale): boxes, exons; UTR, untranslated region; light shaded box, coding region; shaded and dark shaded boxes, nucleotide sequences coding for protein domains (PH: pleckstrin homology domain, RA: ras association domain; RhoGAP: RhoGAP domain).



Schematic representation of ARHGAP20 protein variants as deduced from the transcripts. Hatched box, amino-terminal extension of unknown function; PH: pleckstrin homology domain, RA: ras association domain; RhoGAP: RhoGAP domain.

## Protein

### Description

The amino-terminal region shows significant homology to a pleckstrin homology (PH) domain commonly found in eukaryotic signaling proteins. Adjacent to the PH domain a Ras association (RA) domain is postulated, which is found in proteins involved in GTPase-mediated signaling processes. The central section of the protein contains a RhoGAP domain, which is crucial for the regulation of Rho-like GTPases by Rho GTPase-activating proteins in the course of transmitting diverse intracellular signals.

### Expression

Predominantly expressed in brain, but transcripts were also detected in peripheral blood lymphocytes.

### Localisation

Cytoplasm

### Function

The presence of a RhoGAP domain in combination with PH and RA modules indicates that ARHGAP20 is involved in the regulation of Rho-family GTPases. ARHGAP20 was shown to be activated by Rap1 and to induce inactivation of Rho, resulting in the neurite outgrowth.

### Homology

Mouse: RarhoGAP (RhoGAP having the RA domain), Arhgap20.

Rat: RahoGAP (RhoGAP having the RA domain), Arhgap20.

## Mutations

**Note:** Single nucleotide polymorphism 1785T/C (transcript variant ARHGAP20-1ad, AY496263).

### Germinal

None detected.

### Somatic

In the tumour cells of one case of B-cell chronic lymphocytic leukemia, the missense mutation 2995T>G (S999A; transcript variant ARHGAP20-1ad, AY496263) was found.

## Implicated in

### B-cell chronic lymphocytic leukemia (B-CLL)

**Note:** In the tumour cells of two B-CLL cases, ARHGAP20 was found affected by translocations that rearranged the gene with BRWD3 (Xq21) and a novel gene on 13q14 (unpublished data), respectively. No fusion transcripts were generated. ARHGAP20 transcript expression is significantly upregulated in B-CLL lymphocytes vs. CD19+ control B cells.

#### **t(X;11)(q21;q23)**

##### Disease

B-cell chronic lymphocytic leukemia (B-CLL).

##### Cytogenetics

t(X;11)(q21;q23).

##### Hybrid/Mutated Gene

ARHGAP20 - BRWD3.

##### Abnormal Protein

None detected.

#### **t(11;13)(q23;q14)**

##### Disease

B-cell chronic lymphocytic leukemia (B-CLL).

##### Cytogenetics

t(11;13)(q23;q14).

##### Hybrid/Mutated Gene

ARHGAP20 - novel gene on 13q14 (unpublished data).

##### Abnormal Protein

None detected.

## References

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*This article should be referenced as such:*

Kalla C. ARHGAP20 (Rho GTPase activating protein 20). *Atlas Genet Cytogenet Oncol Haematol.*2007;11(1):1-3.

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