Gene Section
Mini Review

ARHGAP20 (Rho GTPase activating protein 20)
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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).
ARHGAP20 (Rho GTPase activating protein 20)  
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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


This article should be referenced as such: