

## Gene Section

### Mini Review

# BAALC (brain and acute leukemia, cytoplasmic)

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### Identity

**Hugo:** BAALC

**Other names:** FLJ12015; LOC79870

**Location:** 8q22.3

### DNA/RNA

#### Description

The gene spans over 90kb on plus strand; 8 exons; 3 polyadenylation signals in the 3' untranslated region of exon 8.

### Protein

#### Description

Various transcripts: involving exons 1, 6, and 8 (145 amino acids) or 1, and 8 (54 amino acids) in the neuroectoderm; transcripts involving exons 1,5,6,8 or 1,4,5,6,8, or 1,5,6,7,8, or 1,2,6,8, or 1,2,5,6,8 or 1,2,3,6,8 in leukemic cells; altogether, 8 different transcripts of BAALC, giving rise to 5 different proteins

#### Expression

Transcripts involving exons 1, 6, and 8, or 1, and 8 are found in neuroectoderm tissues; BAALC is also expressed in normal early progenitor cells (CD34+) of the hematopoietic tissues: both uncommitted and lineage-committed progenitor cells (lymphoid T- and B-cell progenitors, myeloid and erythroid progenitors); down regulation occurs with cell differentiation; various transcripts are found in leukemic blasts (see above and below); also found expressed in the mesoderm and the muscle in the Mouse.

### Localisation

Cytoplasmic; found adjacent to the cell membrane in the mouse; may be localized to lipid rafts and may play a synaptic role in the rat brain.

### Homology

No homology with other proteins; the ortholog lacks in lower vertebrates.

### Mutations

#### Somatic

Overexpressed in a subset of acute leucemias.

### Implicated in

***1/4 of acute non lymphocytic leukemia (ANLL) cases and 2/3 of acute lymphocytic leukemia (ALL) case have been found to exhibit high BAALC expression; no expression in chronic leukemias (e.g. chronic myeloid leukemia (CML)); expression in blast crisis of CML***

#### Disease

ANLL cases: M0 ANLL, M1, M2 and M4eo cases, rarely M4 or M5 cases; no M3 case so far; association with the more immature blasts; no correlation with age, sex, haemoglobin level or platelets count, or FLT3 phenotype; BAALC expression is associated with a higher WBC count.

#### Prognosis

Poor prognosis is associated with BAALC

overexpression in ANLL cases: at 3 years, only 35-40% of patients with high BAALC expression were alive; independent risk factor; in particular, BAALC overexpression remains a poor prognostic factor in the absence of FLT3 internal tandem duplication.

### Cytogenetics

Patients present with normal karyotypes, with +8, or with various anomalies.

### Glioblastoma

**Note:** BAALC is also upregulated in normal astrocytes.

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