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# Gene Section
## Short Communication

# TNFRSF17 (tumor necrosis factor receptor superfamily, member 17)

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## Abstract

B Cell Maturation Antigen (BCMA) is a transmembrane signaling protein preferentially expressed on plasma cells. Its ligands include B-Cell Activating Factor (BAFF) and A Proliferation Inducing Ligand (APRIL). BCMA is involved in JNK and NF-kB activation pathways that induce B-cell development and autoimmune responses. BCMA has been implicated in autoimmune disorders as well as B-lymphocyte malignancies.

**Keywords**
BCMA, multiple myeloma, immunity, JNK, NF-kB

## Identity

**Other names:** BCM, BCMA, CD269, TNFRSF13A

**HGNC (Hugo):** TNFRSF17

**Location:** 16p13.13

## DNA/RNA

### Description

Spans 3.8 kb; 3 exons, 2 introns.

### Transcription

961 nt mRNA.

## Protein

### Description

184 aa; a glycoprotein with a single transmembrane domain; N-terminally glycosylated at asparagine 42 regulates its function (Huang et al., 2013).
BCMA is a signaling protein that activates the NF-kB.
BCMA contains one cysteine rich domain and a DXL motif, comprised of (Phe/Tyr/Trp)-Asp-Xaa-Leu-(Val/Thr)-(Arg/Gly), to facilitate ligand binding with BAFF or APRIL (Hymowitz et al., 2005).

### Expression

Preferentially expressed in lymph node tissue, B lymphoblast, NK cells, and dendritic cells.

### Localisation

Plasma membrane.

### Function

BCMA mediates the development and survival of B cell lymphocytes that maintain humoral immunity.

### Homology

TNFRSF17 is conserved in P. Troglodytes, M. mulatta, C. lupus, B. taurus, M. musculus, and R. norvegicus.

# Mutations

22 single nucleotide polymorphisms (SNP) have been reported.

# Implicated in

## *Multiple myeloma*

**Disease**

Multiple myeloma is a fatal malignancy that involves the abnormal proliferation and accumulation of plasma cells characterized by their resistance to apoptosis. Sanchez et al. not only showed increased expression of BCMA in patients' plasma cells, but that patients with multiple myeloma have a high concentration of BCMA protein circulating in their blood. Furthermore, the group found that higher levels of circulating BCMA are associated with a worse prognosis.

**Prognosis**

Poor.

## *Acute myelomonocytic leukemia (AMML) with eosinophilia*

**Disease**

AMML is a malignancy involving myeloid precursors of the bone marrow. Although this condition is rare, it is a common type of pediatric acute myeloid leukemia. In about 50% of the cases of AMML, increased numbers of abnormal eosinophils are observed in the bone marrow (eosinophilia). AMML patients with eosinophilia who carry a pericentric inversion at 16(p13q22) tend to have a better prognosis.

**Prognosis**

61% 5 year survival rate; AMML patients who carry the pericentric inversion have a higher rate of complete remission than those without the inversion (Chang et al., 2006).

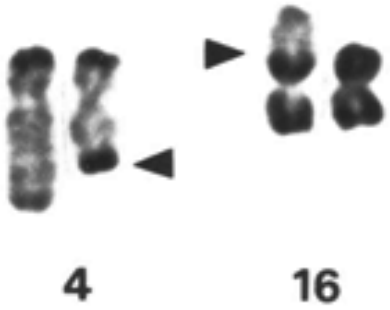
**Cytogenetics**

Pericentric inversion at 16(p13q22) in 3-5% of AMML cases.

## *T cell lymphoma*

**Note**

(4;16)(q26;p13.1) chromosome translocation, which led to the discovery of BCMA, was noted by Laâbi et al. in a single case of T cell lymphoma. This translocation resulted in a IL-2/BCMA composite gene.




**Partial R-Banded karyotype.** The karyotype shows a (4;16) (q26;p13.1) chromosomal translocation that led to the identification of BCMA by Laâbi et al. (Laâbi et al., 1992).

## *Acute monocytic leukemia*

t(8;16)(p11;p13) translocation has been noted in cases of acute monocytic leukemia (Laâbi et al., 1992).

## *Follicular lymphoma*

Translocation (16;18)(p13;q21.3) has been reported in follicular lymphoma (Mahmoodi et al., 2004).

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