# BLNK deficiency

## GENERAL INFORMATION

### Description:

The BLNK deficiency is caused by BLNK gene, a scaffold protein that binds BTK, PLCγ2, Grb2, Vav and Nck and is associated with intracellular calcium mobilization, essential for cell activation. All patients with defects in BLNK have a block in B cell differentiation at the pro-B to pre-B cell transition. Two patients with agammaglobulinemia and defects in BLNK have been identified.

### Alternative names:

- Hypoglobulinemia and absent B cells

### Classification:

- Deficiencies predominantly affecting antibody production
  - Agammaglobulinemia

### Inheritance:

Autosomal recessive

### OMIM:

- #601495 Agammaglobulinemia, non-bruton type
- +604515 B-cell linker protein; BLNK

### Cross references:

**Phenotype related immunodeficiencies:**

- IDR factfile for X-linked agammaglobulinemia
- IDR factfile for X-linked hypogammaglobulinemia with growth hormone deficiency
- IDR factfile for Igα deficiency
- IDR factfile for μ heavy-chain deficiency
- IDR factfile for λ5 surrogate light-chain deficiency

### Incidence:

Incidence is not known.

## CLINICAL INFORMATION

### Description:

Patients with BLNK deficiency have normal growth and development. They have reccurent otitis and episodes of pneumonia and no detectable serum IgG, IgM or IgA, and less than 1% B cells in circulation. After gammaglobulin replacement, patient did well except for chronic otitis and sinusitis, hepatitis C acquiered, and an episode of protein-losing enteropathy.

### Diagnosis:

### Diagnostic laboratories:

**Clinical:**

- Agammaglobulinemia, autosomal recessive, ORPHANET
- Agammaglobulinemia, eMedicine

## Therapeutic options:

- (Intravenous) immunoglobulins, and antibiotic therapy. Oral poliovaccine should not be given because of the risk of paralytic disease.
- Bruton Agammaglobulinemia, eMedicine
- Hypogammaglobulinemia, eMedicine

## Research programs, clinical trials:

- Improved Healthcare for Patients with Primary Antibody Deficiencies through new Strategies Elucidating their Pathophysiology (IMPAD)
- European Initiative for Primary Immunodeficiencies
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

# GENE INFORMATION

## Names:

**HUGO name:** BLNK
**Alias(es):** BLNK-s, Ly57, SLP-65, SLP65, B-cell linker, BASH, BCA

## Localization:

**Reference sequences:**

**DNA:** BLNK_DNA (IDRefSeq) , **cDNA:** AF068180 (EMBL) , **Protein:** O75498 (SWISSPROT)

**Chromosomal Location:**

10q23.2-q23.33

**Maps:**

BLNK (Map View)

## Variations / Mutations:

- BLNKbase; Mutation registry for BLNK deficiency

## Other gene-based resources:

Ensembl: ENSG00000095585, GENATLAS: BLNK, GeneCard: BLNK, UniGene: 444049, Entrez Gene: 29760, euGenes: 29760, GDB: 11504298

# PROTEIN INFORMATION

## Description:

## Other features:

## Expression pattern for human:

| Tissue | Exp. (%) | Clones |
|---|---|---|
| pheochromocytoma | 22.49 | 1:1560 |
| leukocyte | 15.62 | 4:8982 |
| lymph | 9.26 | 17:64395 |
| B-cells | 8.49 | 4:16533 |
| tonsil, enriched for germinal center b-cells | 6.72 | 7:36522 |
| lung metatastic chondrosarcoma | 5.44 | 1:6448 |
| corresponding non cancerous liver tissue | 5.04 | 2:13909 |
| mixed | 4.65 | 8:60341 |
| bone marrow | 3.53 | 2:19854 |
| human skeletal muscle | 3.26 | 1:10746 |

## Animal models:

**Mouse:**

MGD: ; Ly57

**Fly:**

euGenes: ; jp

**C. elegans:**

euGenes: ; T22C1.7

# OTHER RESOURCES

# Societies:

**General:**

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- March of Dimes Birth Defects Foundation
- NIH/National Institute of Allergy and Infectious Diseases
- European Society for Immunodeficiencies