

# Factor B deficiency

## GENERAL INFORMATION

### Description:

Factor B is a serine protease highly homologous to C2. The gene for factor B is part of the HLA class III region on chromosome 6. Factor B is a part of the alternate pathway which plays an important role in natural immunity. Factor B cleaves C3 and C5 and is expressed primarily in hepatocytes and monocytic cells.

### Alternative names:

- Complement factor B deficiency

### Classification:

- Defects of the alternative complement pathway

### Inheritance:

Autosomal recessive

### OMIM:

- \*138470 Properdin factor B; BF

### Cross references:

#### Phenotype related immunodeficiencies:

- IDR factfile for C2 deficiency

### Incidence:

Incidence is not known.

## CLINICAL INFORMATION

### Description:

Patients have meningococemia and a decreased classical pathway activity and no detectable alternative pathway activity. No functional factor B can be detected, and factor B protein has abnormal molecular weight. The Factor B deficiency is associated with severe fulminant pyogenic neisserial infections with a high mortality rate.

### Diagnosis:

#### Diagnostic laboratories:

#### Clinical:

- Complement deficiency, eMedicine

#### Therapeutic options:

- Fresh frozen plasma is used for emergent replacement of complement components. Supportive therapy is used for complement deficiencies. Prophylactic antibiotics for the infections.
- Complement deficiency, eMedicine
- Complement deficiency, eMedicine

#### Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies
- Molecular and Clinical Studies of Primary Immunodeficiency diseases, ClinicalTrials.gov
- Swegene Project

## GENE INFORMATION

### Names:

**HUGO name:** BF

**Alias(es):** B-factor, properdin, Complement factor B precursor (EC 3.4.21.47), C3/C5 convertase, Properdin factor B, Glycine-rich beta glycoprotein, PBF2

### Localization:

#### Reference sequences:

**DNA:** AF019413 (GenBank) , **cDNA:** NM\_001710 (GenBank) , **Protein:** P00751 (SWISSPROT) Other Sequences

#### Chromosomal Location:

6p21.3

#### Maps:

BF (Map View)

### Other gene-based resources:

Ensembl: ENSG00000166285, GENATLAS: BF, GeneCard: BF, UniGene: 69771, Entrez Gene: 629, euGenes: 629, GDB: 119726

## PROTEIN INFORMATION

### Description:

#### Protein function:

Factor B which is part of the alternate pathway of the complement system is cleaved by Factor D into 2 fragments: Ba and Bb. Bb, a serine protease, then combines with complement factor 3b to generate the C3 or C5 convertase. It has also been implicated in proliferation and differentiation of preactivated B lymphocytes, rapid spreading of peripheral blood monocytes, stimulation of lymphocyte blastogenesis and lysis of erythrocytes. Ba inhibits the proliferation of preactivated B lymphocytes.

#### Catalytic activity:

Cleaves C3 in the alpha-chain to yield C3a and C3b. Cleaves C5 in the alpha-chain to yield C5a and C5b. Both cleavages take place at the C-terminal of an Arginine residue.

#### Subunit:

Monomer

#### Miscellaneous:

Factor B is a Major Histocompatibility Complex Class-III protein.

#### Polymorphism:

Two major variants, F and S, and 2 minor variants, as well as at least 14 very rare variants, have been identified.

**Other features:****Other related resources:**

PIR: BBHU, InterPro: IPR001314;  
 Chymotrypsin, InterPro: IPR001254;  
 Ser\_protease\_Try, InterPro: IPR000436;  
 Sushi\_SCR\_CCP, InterPro: IPR002035;  
 VWF\_A, Pfam: PF00084; sushi, Pfam:  
 PF00089; trypsin, Pfam: PF00092; vwa,  
 SMART: SM00032; CCP, SMART: SM00020;  
 Tryp\_SPc, SMART: SM00327; VWA,  
 PROSITE: PS50240; TRYPSIN\_DOM,  
 PROSITE: PS00134; TRYPSIN\_HIS,  
 PROSITE: PS00135; TRYPSIN\_SER,  
 PROSITE: PS50234; VWFA

**Expression pattern for human:**

<b>Tissue</b>	<b>Exp. (%)</b>	<b>Clones</b>
uterus, pooled	15.47	52:15533
liver	8.34	47:26031
ovary, tumor tissue	8.22	2:1125
gall bladder	7.59	4:2435
nasopharynx	7.15	1:646
ovary, pooled	7.12	9:5846
hepatocellular carcinoma	6.82	21:14226
colonic mucosa with ulcerative colitus	3.79	1:1218
placenta human 8 week	3.44	3:4035
corresponding non cancerous liver tissue	2.99	9:13909

**OTHER RESOURCES****Societies:****General:**

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