Mannose-binding lectin deficiency

GENERAL INFORMATION

Description:

Mannose binding lectin protein is a member of collectin family and important for innate immunity. MBL-deficiency has been related to a number of diseases. MBL deficiency mainly results from three mutations in exon 1 of the gene and is associated with both increased susceptibility to infections and autoimmune disease.

Alternative names:

- Mannan-binding protein deficiency

Classification:

- Complement regulatory proteins
  - Mannose-binding lectin deficiency

Inheritance:

Autosomal recessive/Autosomal dominant

OMIM:

- *154545 Lectin, mannose-binding, soluble, 2; MBL2

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for C5 deficiency
- IDR factfile for C6 deficiency
- IDR factfile for C7 deficiency
- IDR factfile for C8 alfa-polypeptide deficiency
- IDR factfile for C8 beta-polypeptide deficiency
- IDR factfile for C8 gamma-polypeptide deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

MBL deficiency is associated with an increased frequency of infection in both adults and children. Many patients have systemic lupus erythematous (SLE).

Diagnosis:

Diagnostic laboratories:

Clinical:

- Complement deficiency, eMedicine

Therapeutic options:

- Fresh frozen plasma is used for emergent replacement of complement components. Prophylactic antibiotics for the infections. Acute infections are treated with appropriate antibiotics and prevented also with vaccination (meningococcal, pneumococcal, and haemophilus).
- 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: MBL2
Alias(es): COLEC1, MBL, MBP1, mannose-binding lectin (protein C) 2, soluble (opsonic defect), Mannose-binding protein C precursor (MBP-C), MBP1, Mannan-binding protein, Mannose-binding lectin

Localization:

Reference sequences:

DNA: X15954 (EMBL) AF080510 (EMBL) Y16576 (EMBL) Y16577 (EMBL) Y16578 (EMBL) Y16579 (EMBL) Y16580 (EMBL) Y16581 (EMBL) Y16582 (EMBL) , **cDNA:** X15422 (EMBL) AF360991 (EMBL) , **Protein:** BAB17020 (GenPept) Other Sequences

Chromosomal Location:

10q11.2-q21

Maps:

MBL2 (Map View)

Other gene-based resources:

Ensembl: ENSG00000165471, GENATLAS: MBL2, GeneCard: MBL2, UniGene: 499674, Entrez Gene: 4153, euGenes: 4153, GDB: 120167

PROTEIN INFORMATION

Description:

Protein function:

Binds mannose and n-acetylglucosamine in a calcium-dependent manner. Is capable of host defense against pathogens, by activating the classical complement pathway independently of the antibody.

Subunit:

Oligomeric complex of 6 set of homotrimers.

Structures (PDB):

1HUP Human mannose-binding protein carbohydrate recognition domain trimerizes through a triple α-helical coiled-coil.

Domains:

Cys-rich domain: 21-41

Collagen-like domain: 42-99

C-type lectin (short form) domain: 153-246

Other features:

Signal peptide : 1-20

Mannose-binding protein c: 21-248

Disulfide bond : 155-244

Disulfide bond : 222-236

Other related resourses:

PIR: LNHUMC, InterPro: IPR008160; Collagen, InterPro: IPR001304; Lectin_C, Pfam: PF00059; lectin_c, Pfam: PF01391; Collagen, SMART: SM00034; CLECT, PROSITE: PS00615; C_TYPE_LECTIN_1, PROSITE: PS50041; C_TYPE_LECTIN_2

Expression pattern for human:

Tissue	Exp. (%)	Clones
corresponding non cancerous liver tissue	69.29	7:13955
liver	18.80	5:36737
hepatocellular carcinoma	9.66	1:14298
pool, liver+spleen	2.24	1:61534

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