Factor D deficiency

GENERAL INFORMATION

Description:

Factor D is part of the alternative complement pathway and is identical to adipisin. It is synthesized in adypocytes, muscle cells, lung, and monocytes/macrophages. Factor D converts factor B to its end product.

Alternative names:

- Complement factor D deficiency

Classification:

- Defects of the alternative complement pathway

Inheritance:

Autosomal recessive

OMIM:

- *134350 Factor D

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for C4 binding protein α deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have recurrent sinopulmonary infections or recurrent systemic meningococcal infections.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Complement deficiency, eMedicine

Therapeutic options:

- Fresh frozen plasma is used for emergent replacement of complements 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: CFD
Alias(es): D component of complement (adipsin), Complement factor D precursor, C3 convertase activator, Properdin factor D, Adipsin, DF

Localization:

Reference sequences:

cDNA: M84526 (EMBL) , **Protein:** P00746 (SWISSPROT)

Chromosomal Location:

19p13.3

Maps:

DF (Map View)

Other gene-based resources:

Ensembl: ENSG00000197766, GENATLAS: DF, GeneCard: DF, UniGene: 155597, Entrez Gene: 1675, euGenes: 1675, GDB: 132645

PROTEIN INFORMATION

Description:

Protein function:

Factor D cleaves factor B when the latter is complexed with factor C3b, activating the C3bb complex, which then becomes the C3 convertase of the alternate pathway. Its function is homologous to that of C1s in the classical pathway.

Catalytic activity:

Cleaves component factor B (arg-|-lys) when in complex with C3b or with cobra venom factor (CVF).

Structures (PDB):

1DFP	Factor D Inhibited By Diisopropyl Fluorophosphate
1DST	Mutant Of Factor D With Enhanced Catalytic Activity
1DSU	Human Factor D, Complement Activating Enzyme

Other features:

Signal peptide : 1-20

Propeptide Activation peptide: 21-25

Complement factor d: 26-253

Disulfide bond : 51-67

Disulfide bond : 148-214

Disulfide bond : 179-195

Disulfide bond : 204-229

Other related resourses:

PIR: DBHU, InterPro: IPR001314; Chymotrypsin, InterPro: IPR001254; Ser_protease_Try, Pfam: PF00089; trypsin, SMART: SM00020; Tryp_SPc, PROSITE: PS50240; TRYPSIN_DOM, PROSITE: PS00134; TRYPSIN_HIS, PROSITE: PS00135; TRYPSIN_SER

Expression pattern for human:

Tissue	Exp. (%)	Clones
adipose, white adipose	33.11	1:268
sciatic nerve	10.11	1:878
pool, placenta	6.77	9:11805
subchondral bone	6.62	1:1340
gall bladder	3.63	1:2445
grade-2-chondrosarcoma	3.36	1:2641
lung metatastic chondrosarcoma	2.75	2:6458
prostate	2.56	22:76215
muscle, pectoral muscle	2.37	4:14979
genitourinary tract	2.32	1:3824

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