Factor I deficiency

GENERAL INFORMATION

Description:

Factor I consists of two disulfide-linked polypetides which regulate the alternative pathway-cleaving enzyme. The alternative pathway in uncontrolled in the patients resulting in continous activation and cleavage of native C3 throgh the alternative pathway and the production of C3b. Patients with factor I deficiency have a secondary deficiency of C3 with markedly reduced levels of C3 in their serum. Total hemolytic activity of the serum is very low or undetectable. Level of factor H, factor B, and properdin is reduced.

Alternative names:

- Complement component I deficiency

Classification:

- Defects of complement regulatory proteins

Inheritance:

Autosomal recessive

OMIM:

- +217030 I factor; IF

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for C4 binding protein α deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have an increased susceptibility to infections. Neisserial infections have been reported in half of the cases. Due to defective opsonization, patients are susceptible to recurrent pyogenic infections. Some patients also can be asymptomatic. Rheumatic disorder are not common.

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.
- 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, South Western

GENE INFORMATION

Names:

HUGO name: CFI
Alias(es): C3b inactivator, I factor complement, complement component I, Complement factor I precursor, C3B/C4B inactivator

Localization:

Reference sequences:

DNA: IF_DNA (IDRefSeq) , **cDNA:** J02770 (EMBL) , **Protein:** P05156 (SWISSPROT) Other Sequences

Chromosomal Location:

4q25

Maps:

IF (Map View)

Variations / Mutations:

- CFIbase; Mutation registry for Factor I deficiency

Other gene-based resources:

Ensembl: ENSP00000265172, GENATLAS: IF, GeneCard: IF, UniGene: 312485, Entrez Gene: 3426, euGenes: 3426, GDB: 120077

PROTEIN INFORMATION

Description:

Protein function:

Responsible for cleaving the alpha-chains of C4b and C3b in the presence of the cofactors C4-binding protein and Factor H respectively.

Catalytic activity:

Inactivates complement subcomponents C3b, IC3b and C4b by proteolytic cleavage.

Subunit:

Heterodimer of a light and heavy chains linked by disulfide bonds.

Subcellular location:

Extracellular.

Other features:

Other related resourses:

PIR: A29154, InterPro: IPR001314; Chymotrypsin, InterPro: IPR003884; FacI_MAC, InterPro: IPR002172; LDL_recept_A, InterPro: IPR001254; Ser_protease_Try, InterPro: IPR001190; Srcr_receptor, Pfam: PF00057; ldl_recept_a, Pfam: PF00089; trypsin, Pfam: PF00530; SRCR, SMART: SM00057; FIMAC, SMART: SM00192; LDLa, SMART: SM00202; SR, SMART: SM00020; Tryp_SPc, PROSITE: PS01209; LDLRA_1, PROSITE: PS50068; LDLRA_2, PROSITE: PS00420; SRCR_1, PROSITE: PS50287; SRCR_2, PROSITE: PS50240; TRYPSIN_DOM, PROSITE: PS00134; TRYPSIN_HIS, PROSITE: PS00135; TRYPSIN_SER

Expression pattern for human:

Tissue	Exp. (%)	Clones
ovary, tumor tissue	13.63	1:1125
thyroid gland	13.50	1:1136
muscle, leg skeletal muscle	8.98	1:1708
liver	7.66	13:26031
T cells from T cell leukemia	6.40	1:2397
uterus, pooled	4.94	5:15533
pancreas, islet	4.23	1:3625
kidney, pooled	4.14	2:7404
B cells germinal	4.07	2:7537
genitourinary tract	4.02	1:3813

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