Properdin factor C deficiency

GENERAL INFORMATION

Description:

Properdin is synthesized by monocytes, hepatocytes, and T cells, and it is a component of the secondary granule in neutrophiles. In some families, properdin is undetectable, while in other affected males may have levels as high as 10% of normal. In other kindreds the serum level of properdin are normal but the protein is dysfunctional. Three phenotypes have been reported: complete deficiency (type I), incomplete deficiency (type II), and dysfunction of properdin (type III).

Alternative names:

- Properdin P factor deficiency

Classification:

- Defects of the alternative complement pathway

Inheritance:

X-linked

OMIM:

- #312060 Properdin deficiency, X-linked
- *300383 Properdin P Factor, Complement PFC

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for C4 binding protein α deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have higher susceptibility to bacterial infections; especially to meningococcal infections. More than half of patients with properdin deficiency have had meningococcal disease. Patients with properdin deficiency have an intact classical pathway and normal levels of serum C3 and other components of the alternative pathway are present in normal levels. SLE (systemic lupus erythematous) and discoid lupus are possible.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Complement deficiency, eMedicine
- Properdin deficiency, ORPHANET

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:

GENE INFORMATION

Names:

HUGO name: CFP
Alias(es): BFD, PFC, PFD, properdin P factor, complement, Properdin precursor , Factor P

Localization:

Reference sequences:

DNA: X70872 (EMBL) , **cDNA:** NM_002621 (GenBank) , **Protein:** P27918 (SWISSPROT) Other Sequences

Chromosomal Location:

Xp11.3-p11.23

Maps:

PFC (Map View)

Variations / Mutations:

- CFPbase; Mutation registry for properdin deficiency

Other gene-based resources:

Ensembl: ENSG00000126759, GENATLAS: PFC, GeneCard: PFC, UniGene: 53155, Entrez Gene: 5199, euGenes: 5199, GDB: 120275

PROTEIN INFORMATION

Description:

Protein function:

A positive regulator of the alternate pathway of complement. It binds to and stabilizes the C3- and C5-convertase enzyme complexes.

Other features:

Other related resourses:

PIR: S29126, InterPro: IPR000884; TSP1, Pfam: PF00090; tsp_1, SMART: SM00209; TSP1, PROSITE: PS50092; TSP1

Expression pattern for human:

Tissue	Exp. (%)	Clones
adipose, white adipose tissue	27.20	1:987
whole blood	21.96	2:2445
spleen	11.14	3:7229
mixed	6.67	15:60341
ovary (pool of 3)	6.13	1:4380
leukocyte	5.98	2:8982
aorta	5.23	2:10275
pituitary	4.53	1:5926
muscle, pectoral muscle	3.59	2:14965
B-cells	3.25	2:16533

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