

C6 deficiency

GENERAL INFORMATION

Description:

The most common form of C6 deficiency is characterised by undetectable levels of C6. Subtotal deficiency of C6(C6SD) is characterised by 1%-2% of the normal serum antigenic levels of C6. Subtotal deficiency of C6 has also been seen in association with subtotal C7 deficiency.

Alternative names:

- Complement component 6 deficiency

Classification:

- Defects of the classical complement cascade proteins

Inheritance:

Autosomal recessive

OMIM:

- +217050 Complement component 6 deficiency

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for C5 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
- IDR factfile for C9 deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients with C6 deficiency lack serum bactericidal activity and most commonly present with systemic blood-born meningococcal infections. Disseminated gonococcal infections have rarely been described. Few patients with C6 deficiencies present systemic lupus erythematosus(SLE).

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. Autoimmune disease is treated in the normal way.
- 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: C6

Alias(es): complement component 6,
Complement component C6 precursor

Localization:

Reference sequences:

DNA: C6_DNA (C6base) , **cDNA:** NM_000065
(GenBank) , **Protein:** P13671 (SWISSPROT)
Other Sequences

Chromosomal Location:

5p13

Maps:

C6 (Map View)

Variations / Mutations:

- C6base; Mutation registry for C6 deficiency.

Other gene-based resources:

Ensembl: ENSG00000039537, GENATLAS: C6,
GeneCard: C6, UniGene: 481992, Entrez Gene:
729, euGenes: 729, GDB: 119045

PROTEIN INFORMATION

Description:

Protein function:

Involved in the formation of the lytic C5b-9m complex.

Subcellular location:

Secreted

Post-translational modification:

All cysteine residues are assumed to be cross-linked to one another. Individual modules containing an even number of conserved cysteine residues are supposed to have disulfide linkages only within the same module.

Polymorphism:

The sequence shown is that of allotype C6a.

Other features:

Other related resources:

PIR: A34372, InterPro: IPR003884; FacI_MAC, InterPro: IPR002172; LDL_recept_A, InterPro: IPR001862; MAC_perforin, InterPro: IPR000436; Sushi_SCR_CCP, InterPro: IPR000884; TSP1, InterPro: IPR002350; kazal, Pfam: PF00050; kazal, Pfam: PF00057; ldl_recept_a, Pfam: PF00084; sushi, Pfam: PF00090; tsp_1, Pfam: PF01823; MACPF, SMART: SM00032; CCP, SMART: SM00057; FIMAC, SMART: SM00280; KAZAL, SMART: SM00192; LDLa, SMART: SM00457; MACPF, SMART: SM00209; TSP1, PROSITE: PS00022; EGF_1, PROSITE: PS01186; EGF_2, PROSITE: PS01209; LDLRA_1, PROSITE: PS50068; LDLRA_2, PROSITE: PS00279; MAC_PERFORIN, PROSITE: PS50092; TSP1

Expression pattern for human:

Tissue	Exp. (%)	Clones
corresponding non cancerous liver tissue	22.65	11:13909
hepatocellular carcinoma	20.13	10:14226
gall bladder	11.76	1:2435
liver	9.90	9:26031
human lung epithelial cell lines untreated lps 6hr to lps	9.12	2:6278
pool, liver+spleen	7.94	17:61327
ovary (pool of 3)	6.54	1:4380
pancreas, exocrine	4.01	3:21418
kidney, pooled	3.87	1:7404
germ cell	1.43	1:20077

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