C7 deficiency

GENERAL INFORMATION

Description:

Patients with C7 deficiency have markedly diminished total hemolytic complement activity and little if any C7 in their serum. Serum bactericidal activity is markedly reduced and is responsible for the increased risk of neiserrial infections in C7-deficient patients. In the second type of C7 deficiency, the quantity of C7 is diminished and the protein exhibits an altered isoelectric point. C7 deficiency has been in association with subtotal deficiency of C6.

Alternative names:

- C7 deficiency
- Complement component 7 deficiency

Classification:

- Defects of the classical complement cascade proteins

Inheritance:

Autosomal recessive

OMIM:

- +217070 Complement component 7 deficiency

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for C5 deficiency
- IDR factfile for C6 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:

Systemic neisserial infections occur in 60% of reported cases of C7-deficient patients. Other manifestation include SLE, rheumatoid arthritis, pyoderma gangrenosum, and scleroderma.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Complement deficiency, eMedicine

Genetic:

- EDDNAL

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. Specific treatment of autoimmune disease is needed.
- 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: C7
Alias(es): complement component 7, Complement component C7 precursor

Localization:

Reference sequences:

DNA: C7_DNA (C7base) , **cDNA:** J03507 (EMBL) , **Protein:** P10643 (SWISSPROT) Other Sequences

Chromosomal Location:

5p13

Maps:

C7 (Map View)

Variations / Mutations:

- C7base; Mutation registry for C7 deficiency.

Other gene-based resources:

Ensembl: ENSG00000112936, GENATLAS: C7, GeneCard: C7, UniGene: 78065, Entrez Gene: 730, euGenes: 730, GDB: 119046

PROTEIN INFORMATION

Description:

Protein function:

C7 is a constituent of the membrane attack complex. C7 binds to C5b forming the C5b-7 complex, where it serves as a membrane anchor.

Subunit:

Monomer or dimer; as a C5b-7 complex it can also form multimeric rosettes.

Post-translational modification:

C7 has 28 disulfide bridges.

Other features:

Other related resourses:

PIR: A27340, InterPro: IPR003884; FacI_MAC, InterPro: IPR002172; LDL_recept_A, InterPro: IPR001862; MAC_perforin, InterPro: IPR000436; Sushi_SCR_CCP, InterPro: IPR000884; TSP1, Pfam: PF00057; ldl_recept_a, Pfam: PF00084; sushi, Pfam: PF00090; tsp_1, Pfam: PF01823; MACPF, SMART: SM00032; CCP, SMART: SM00057; FIMAC, 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
esophageal squamous cell carcinoma	96.12	1:5
ovary, tumor tissue	0.85	2:1125
adrenal cortico adenoma for cushing's syndrome	0.46	1:1050
thyroid gland	0.42	1:1136
normal head/neck tissue	0.37	1:1292
adrenal gland	0.28	10:17391
ear, cochlea	0.16	3:9125
genitourinary tract	0.13	1:3813
prostate, epithelium	0.10	2:9299
kidney	0.09	23:117548

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