C9 deficiency

GENERAL INFORMATION

Description:

C9 is a glycoprotein wich has sequence homology to C8alfa, C8beta, C6, and perforin. C9 is produced by hepatocytes, it is cytokine inducible and behaves as an acute phase reactant. Patients with C9 deficiency have reduced levels of C9 in their serum. They possess some serum hemolytic and bactericidal activity but less than in normal serum.

Alternative names:

- Complement component 9 deficiency

Classification:

- Defects of the classical complement cascade proteins

Inheritance:

Autosomal recessive

OMIM:

- +120940 Complement component 9; C9

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for C5 deficiency
- IDR factfile for C6 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

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients with C9 deficiency are often asymptomatic but some present systemic meningococcal infections. In Japan C9 deficiency is relatively common. Patients with C9 deficiency have the ability to kill Neisseria but at a slower rate.

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. Specific treatment of autoimmune disease is followed.
- 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: C9
Alias(es): complement component 9, Complement component C9 precursor

Localization:

Reference sequences:

DNA: C9_DNA (C9base) , **cDNA:** X02176 (EMBL) , **Protein:** P02748 (SWISSPROT) Other Sequences

Chromosomal Location:

5p13

Maps:

C9 (Map View)

Variations / Mutations:

- C9base; Mutation registry for C9 deficiency.

Other gene-based resources:

Ensembl: ENSG00000113600, GENATLAS: C9, GeneCard: C9, UniGene: 1290, Entrez Gene: 735, euGenes: 735, GDB: 119738

PROTEIN INFORMATION

Description:

Protein function:

C9 is the final component of the complement system to be added in the assembly of the membrane attack complex. It is able to enter lipid bilayers, forming transmembrane channels.

Post-translational modification:

Thrombin cleaves factor C9 to produce C9a and C9b.

Other features:

Other related resourses:

PIR: C9HU, InterPro: IPR006210; EGF-like, InterPro: IPR002172; LDL_recept_A, InterPro: IPR001862; MAC_perforin, InterPro: IPR000884; TSP1, Pfam: PF00057; ldl_recept_a, Pfam: PF00090; tsp_1, Pfam: PF01823; MACPF, 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
liver	34.57	34:26031
corresponding non cancerous liver tissue	26.64	14:13909
hepatocellular carcinoma	22.33	12:14226
placenta human 8 week	13.12	2:4035
muscle (skeletal)	2.77	1:9571
pool, lung+testis+b-cell	0.48	1:55714
brain	0.10	1:274929

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