Hereditary angioedema

GENERAL INFORMATION

Description:

Hereditary angioedema is episodic and asymptomatic in most patients. There are 2 types of the disorder. In type I, representing 85% of patients, serum levels of C1NH are less than 35%. In type II, the levels are normal or elevated, but the protein is nonfunctional. The two types are clinically similar.

Alternative names:

- HANE
- HAE
- Angioneurotic edema, hereditary
- C1 esterase inhibitor deficiency
- C1 inhibitor deficiency
- Hereditary angioedema type I and type II
- Quinke's disease

Classification:

- Defects of complement regulatory proteins
  - Hereditary angioedema

Inheritance:

Autosomal dominant

OMIM:

- #106100 Angioedema, hereditary; HAE
- *606860 Angioedema, hereditary; HAE

Cross references:

Incidence:

1:50,000-150,000

CLINICAL INFORMATION

Description:

Patients can develop rapid swelling of the hands, feet, limbs, face, intestinal tract, or airway (larynx or trachea). HANE is characterized by recurrent episodes of angioedema involving any part of the body. Laryngeal edema is common, and it is the major cause of death. Angioedema of the gastrointestinal tract may frequently mimic an acute abdomen.

Diagnosis:

Diagnostic laboratories:

Clinical:

- GeneTest
- ORPHANET
- eMedicine

Genetic:

- Laboratorio di Genetica Medica - Policlinico Universitario Tor Vergata (Roma), EDDNAL
- DNA Diagnostics Laboratory - Department of Human Genetics (Nijmegen), EDDNAL

Therapeutic options:

- C1 inhibitor concentrate is preferred for acute treatment. Androgens such as winstrol, danazol, and oxandrolone for possible prevention of episodes. Hypotension accompanies abdominal attacks in some patients and fluid replacement therapy is required. A combination of Demerol and Compazine suppositories, and possibly Dicyclomine is useful to relieve abdominal pain and vomiting.
- Angioedema, Hereditary, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies.
- Study of Heparin Prophylaxis of Hereditary Angioedema Exacerbations, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: SERPING1
Alias(es): C1-INH, C1IN, C1NH, complement component 1 inhibitor (angioedema, hereditary), Plasma protease C1 inhibitor precursor (C1 Inh) (C1Inh)

Localization:

Reference sequences:

DNA: AF435921 (GenBank) , **cDNA:** NM_000062 (GenBank) , **Protein:** P05155 (SWISSPROT) Other Sequences

Chromosomal Location:

11q12-q13.1

Maps:

SERPING1 (Map View)

Other gene-based resources:

Ensembl: ENSG00000149131, GENATLAS: SERPING1, GeneCard: SERPING1, UniGene: 384598, Entrez Gene: 710, euGenes: 710, GDB: 119041

PROTEIN INFORMATION

Description:

Protein function:

Activation of the C1 complex is under control of the C1-inhibitor. It forms a proteolytically inactive stoichiometric complex with the C1r or C1s proteases. May play a potentially crucial role in regulating important physiological pathways including complement activation, blood coagulation, fibrinolysis and the generation of kinins.

Subcellular location:

Secreted

Post-translational modification:

Highly glycosylated (49%).

Polymorphism:

There are two alleles.

Other features:

Other related resourses:

PIR: ITHUC1, InterPro: IPR000215; Serpin, Pfam: PF00079; serpin, SMART: SM00093; SERPIN, PROSITE: PS00284; SERPIN

Expression pattern for human:

Tissue	Exp. (%)	Clones
adipose, white adipose	7.78	1:268
nose, olfactory epithelium	5.59	3:1119
dorsal root ganglia	5.57	4:1496
human eye anterior segment	5.26	5:1981
testis, epididymus	4.41	2:945
adipose, white adipose tissue	4.21	2:990
uterus, endometrium	3.48	3:1796
brain, miningioma	2.94	1:709
pheochromocytoma	2.66	2:1569
prostate, broad spectrum prostate tumors	2.54	1:821

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

Disease specific:

- International Patient Organisation for C1 Inhibitor Deficiencies
- United States Hereditary Angioedema Association
- Italian Hereditary Angioedema Association
- Argentinian Hereditary Angioedema Association
- Canadian Hereditary Angioedema Society
- Danish Hereditary Angioedema Association
- French Hereditary Angioedema Association
- German Hereditary Angioedema Association
- Netherlands Hereditary Angioedema Association
- Spanish Association Hereditary Angioedema

Other information sources:

- Immunodeficiencies AND hereditary angioedema