



## Angioedema, Hereditary

### Alternative Names

HAE  
Angioneurotic Edema, Hereditary  
HANE  
C1 Esterase Inhibitor, Deficiency of  
Angioedema, Hereditary, Type I  
Angioedema, Hereditary, Type II

### WHO International Classification of Diseases

Injury, poisoning and certain other consequences of external causes

### OMIM Number

106100

### Mode of Inheritance

Autosomal Dominant

### Gene Map Locus

11q11-q13.1

### Description

Hereditary angioedema is an autosomal dominant inherited abnormality of the immune system, characterized clinically by recurrent episodes of subcutaneous, intestinal, and laryngeal edema. In some cases the laryngeal edema may be so severe as to occlude the upper airway and threaten life. Factors that trigger episodes vary and symptoms typically last 48 to 72 hours, but they can last 4 hours to 1 week.

The disease is a result from deficiency of the plasma protein C1 inhibitor. Two distinct categories of hereditary angioedema are now recognized. Type I affects approximately 85% of all patients with hereditary angioedema, and is characterized by low antigenic and functional levels of C1 inhibitor. Type II is found in approximately 15% of patients with hereditary angioedema, and is defined by normal or elevated levels of C1 inhibitor with low functional activity caused by the secretion of a dysfunctional protein. The prevalence of the

disease has been estimated at 1/50 000, with no reported bias in different ethnic groups.

### Molecular Genetics

Hereditary angioedema is caused by mutation in the C1 inhibitor gene, mapping to chromosome 11q12-q13.1 and comprising eight exons. Rare mutations causing deficiency or dysfunction of C1 inhibitor have been identified throughout the entire length of the gene, as well as common sequence variations of unknown significance. Moreover, the C1 inhibitor locus has clusters of intragenic Alu repeats that predispose to deleterious gene rearrangements. Such gross alterations in the C1 inhibitor gene have been reported in up to 20% of individuals with type I hereditary angioedema. The spontaneous mutation rate is about 25% and more than 100 different C1 inhibitor gene mutations have been described.

### Epidemiology in the Arab World

#### United Arab Emirates

Gomaa (1985) reported a case of hereditary angioedema in a 27 years old female, admitted to the hospital with a two day history of severe abdominal pains accompanied by vomiting and fluid diarrhoea. The patient had previously, in 1975, been diagnosed with hereditary angioedema, and had since been experiencing episodic abdominal pains and occasional swelling of the hands. There had been no history of laryngeal oedema. Clinical investigations showed dehydration, coated tongue and vague tenderness in the abdomen. Laboratory findings revealed low level of C1 esterase inhibitor.

### References

Gomaa MH. Hereditary Angio-oedema. Emirates Med J 1985; 3:137-8.



**Contributors**

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