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MEETING ABSTRACT Open Access

A nationwide survey of hereditary angioedema due to C1 inhibitor deficiency in Italy

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Background

Hereditary angioedema due to C1-inhibitor deficiency (C1-INH-HAE type I) or dysfunction (C1-INH-HAE type II) is a rare disease characterized by recurrent episodes of edema with an estimated frequency of 1:50,000 in the global population without racial or gender differences. In this study we present the results of a nationwide survey of C1-INH-HAE patients referring to 17 Italian centers, the Italian network for C1-INH-HAE, ITACA

Methods

Italian patients diagnosed with C1-INH-HAE from 1973 to 2013 were included in the study. Diagnosis of C1-INH-HAE was based on family and/or personal history of recurrent angioedema without urticaria and on antigenic and/or functional C1-INH deficiency.

Results

983 patients (53% female) from 376 unrelated families were included in this survey. Since 1973, 63 (6%) patients diagnosed with C1-INH-HAE died and data from 3 patients were missing when analysis was performed. Accordingly, the minimum prevalence of HAE in Italy in 2013 is 920:59,394,000 inhabitants, equivalent to 1:64,935. Compared to the general population, patients are less represented in the early and late decades of life: men start reducing after the 5th decade and women after the 6th. Median age of patients is 45 (IQ 28-57), median age at diagnosis is 26 years (IQ 13-41). C1-INH-HAE type 1 are 87%, with median age at diagnosis of 25 (13-40); type 2 are 13% with median age at diagnosis of 31 (IQ 16-49). Functional C1INH is ≤50% in 99% of patients. Antigen C1INH is ≤50% in 99% of type 1. C4 is ≤50% in 96% of patients.

Conclusions

This nationwide survey of C1-INH-HAE provides for Italy a prevalence of 1: 64,935. C1-INH-HAE patients listed in our database seem to have a shorter life expectancy than the general population. Since angioedema symptoms usually start during puberty, the estimated delay in diagnosis is higher than 10 years. The chance of having C1-INH-HAE with C4 plasma levels >50% is < 0.05.

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