Prevalence and Clinical Manifestations of Hereditary Angioedema in Blood Relatives of the Hereditary Angioedema Patients in a City of Düzce Province, Yiğilca, Turkey

Abstract

Introduction: Hereditary angioedema (HAE) is a rare autosomal-dominant disease described by recurring attacks of nonpruritic, nonpitting edema attributable to an inherited deficiency or dysfunction of C1 esterase inhibitor (C1 INH). The aim of this study was to determine the prevalence rate and clinical manifestations of HAE in untested blood relatives of HAE patients in a city of Düzce province, Turkey.

The patients and methods: Overall, 3 patients diagnosed with HAE and their 65 blood relatives (36 female+32 male) were enrolled in the study. Mean age of these subjects was 30 years. Patients with a confirmed diagnosis of HAE recruited blood relatives who had not been evaluated for HAE. Enrolled subjects underwent complement testing (C1 INH antigen, C1 INH function and C4). If the laboratory tests were abnormal, the enrolled subjects were evaluated with a questionnaire for clinical manifestations and begun to be followed up.

Results: In this study group of blood relatives, C1 INH antigen was detected to be low in 26/65 (%40). Only 2/65 (%3) subjects had low C1 INH functional levels. C4 levels were found to be low in 14 of 27 (%52) HAE patients. Of 65 enrolled relatives, 37 (57%) had laboratory test results that ruled out a diagnosis of HAE, 4(6%) were categorized as "HAE not ruled out," and 24(37%) were newly diagnosed with HAE. Of 24 newly diagnosed subjects, nine (38%) reported having experienced symptoms that may have been related to HAE, such as swelling in the face, or extremities or abdominal pain. Median age of 9 symptomatic patients was 48 years, whereas newly diagnosed asymptomatic subjects had a median chronological age of 32 years. These 9 symptomatic patients reported a few swelling episodes per year persisting a couple of days. Swelling in extremities was observed in 7 symptomatic patients, face (lip) swelling in 2 patients, testicular swelling in 1 and abdominal pain was reported in 2 of HAE patients.

Conclusion: This screening study's results underline the importance of testing untested family members and blood relatives of HAE patients to discover this hereditary condition.

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