Proceeding

Prevalence rate and clinical manifestations of hereditary angioedema in untested blood relatives of the hereditary angioedema patients in a turkish city

Abstract

Background: 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). Symptoms can present years before a precise diagnosis is made.

Objective: 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.

Patients and methods: Overall, 3 patients with HAE and 65 blood relatives were enrolled in the study. Mean age of enrolled subjects (36 female+32 male) 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 (C4, C1 INH antigen, and functional C1 INH). If the laboratory tests were abnormal, the enrolled subjects were evaluated with a questionnaire for clinical manifestations and begun to be followed up.

Results: C4 levels were found to be low in 14 of 27 (%52) HAE patients. 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 activity levels of C1 INH. 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 emphasize the importance of testing family members and blood relatives of patients with HAE to identify this hereditary condition.

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Öner Özdemir, Bahri Elmas
Department of Pediatrics, Research and Training Hospital of Sakarya University, Turkey

Correspondence: Öner Özdemir, Division of Allergy and Immunology, Department of Pediatrics, Research and Training Hospital of Sakarya University, Faculty of Medicine, Sakarya University, Adnan Menderes Cad, Sa□lık Sok No: 195, Adapazari, Sakarya, Turkey, Tel 90-264-444-54-00, Fax 90-264-275-91-92, Email ozdemir_oner@hotmail.com

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Conflicts of interest

Authors declare that there is no conflict of interest.

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