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Clinical profile of hereditary angioedema: our experience over 26 years in North India

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Background: Hereditary angioedema (HAE) is a rare genetic disorder. There is limited data in the literature on patients with HAE from developing countries where patient awareness is low and first line medications are not available.

Objective: This study was carried out to analyze the clinical manifestations and laboratory features of patients diagnosed with HAE between January 1996 and April 2022 in a tertiary care center in North India.

Methods: Data of patients with HAE were retrieved from medical records of patients registered in the Paediatric Immunodeficiency Clinic, Postgraduate Institute of Medical Education and Research, Chandigarh, India.

Results: In this study, we included 145 patients (85 male and 60 female) from 53 families who were diagnosed to have HAE. The median age at onset of symptoms was 14 years (range 1-40 years) and the median age at diagnosis was 27 years (range 2-80 years) with a median delay in diagnosis of 11 years (range 0-44 years). Family history was present in 124/145 (85.5%). The mean frequency of exacerbation was 1.6 attacks/month. Swelling over the face (eyelids and/or lips) was the commonest presentation 93.7% (136/145) followed by extremities and genitalia (Table-1). Laryngeal edema was seen in 45.5% (66/145) cases, 17 of them having multiple attacks. Abdominal symptoms were noted in 62.1% of patients, only 1 patient presented with the acute surgical abdomen and underwent exploratory laparotomy. No patient in this study had central nervous system complaints. Mean duration of follow-up is 40.2 months (total- 5829 patient-months). Type I HAE was seen in 119 patients and 18 patients had type-II HAE. Normal C1 INH HAE was seen in 8. Mean serum C4 level and C1 INH were 10.4 mg/dL (Normal-16.7-38.5 mg/dL) and 10.8 mg/dL (Normal-19-37 mg/dL) respectively. Abdominal symptoms and males were significantly more in type-I HAE whereas laryngeal edema and tongue swelling were higher in type-II HAE(Table-2). Due to a lack of first-line medications, patients from our cohort received fresh frozen plasma as emergent therapy and tranexamic acid and stanozolol as prophylactic agents.

Conclusion: This is one of the largest cohorts of HAE from a developing country and shows a median delay of 11 years for diagnosis of the disease. Hence patient awareness about this rare disease is required.

Table 1: Clinical features of HAE patients

Clinical features	Number of patients (n=145)
Asymptomatic	2 (1.3%)
Swelling over face (lips and/or eyelids)	136 (93.7%)
Swelling of extremities	116 (80%)
Swelling of genitalia	15 (10.3%)
Laryngeal edema	66 (45.5%)
Abdominal pain	90 (62.1%)
Tongue swelling	40 (27.5%)
Family history	124/145 (85.5%)
Low serum C4 (Normal- 16.7-38.5 mg/dL)	137 (94.5%)
Low serum C1 INH (Normal-19-37 mg/dL)	119 (82.1%)
Normal C1 INH HAE	8 (5.5%)

Table 2: Comparison between normal and low serum C 1 INH levels:

Characteristics	Type-I HAE (n=119)	Type-II HAE (n=18)	p value
Male	76	4	0.001
Frequency of attacks (per month)	1.6	2	
Swelling of genitalia	11	3	0.275
Laryngeal edema	49	15	0.008
Abdominal swelling	84	4	0.001
Tongue swelling	28	10	0.0073