HAE with normal C1-INH: Treatment and attack frequency based on data from the Canadian 2020 national survey

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Introduction: Hereditary angioedema with normal C1-esterase inhibitor (HAEnC1-INH) is associated with mutations in Factor XII and other genes. In the absence of genetic testing, diagnosis is based on clinical symptoms of angioedema and normal C1-INH functional levels so medication access can be difficult and available medications may not be fully effective.

Methods: In 2020, an online survey was sent to all members of HAE Canada. Survey results were collated and sorted by self-reported HAEnC1-INH and Type 1/2 HAE. Demography, medication use and angioedema attacks are reported and expressed as percent of respondents.

Results: Forty-five adult patients (84% female) with HAEnC1-INH and 106 (76% female) with Type 1/2 HAE responded to the survey. For the prior year, more respondents with HAEnC1-INH reported ≥12 angioedema attacks (50% vs27%) and fewer reported having none (10% vs 21%) compared to those with Type 1/2 HAE. Laryngeal attacks were experienced by 43% with HAEnC1-INH versus 24% with Type 1/2 HAE.

Most respondents indicated that their primary treatment for acute attacks (HAEnC1-INH 41%, Type 1/2 HAE 59%) and prophylaxis (HAEnC1-INH 45%, Type 1/2 HAE 59%) was plasma-derived (pd) C1-INH. Others used icatibant to treat acute attacks (HAEnC1-INH 30%, Type 1/2 HAE 23%).

Discussion: These results suggest pdC1-INH is used to treat a large proportion of patients with HAEnC1-INH despite presumed normal C1 inhibitor function. Half experience >12 attacks per year and 43% have laryngeal attacks - higher than those with HAE Type 1/2. In Canada, new treatments targeting angioedema, genetic testing and precision medicine are needed for HAEnC1-INH patients.

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