



## Educational Activities Significantly Improved Diagnosis of the Rare Disease Hereditary Angioedema

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### ABSTRACT

This study demonstrates the effectiveness of educational activities in assisting the diagnosis of rare conditions, resulting in reduced mortality. Similar programs should be initiated for other rare diseases to reduce misdiagnosis and thereby improve patient care.

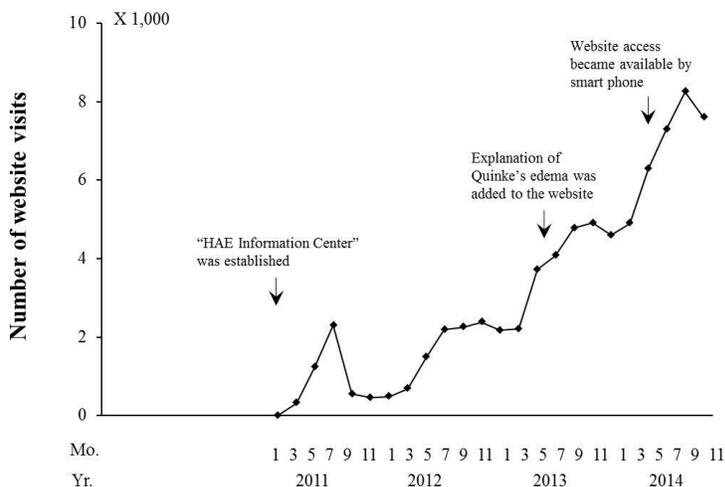
### INTRODUCTION

Hereditary angioedema (HAE) is primarily caused by abnormally low levels of C1 esterase inhibitor and a reduction in function, which inhibits the complement system in the blood. It is estimated that HAE affects between 1 in 10,000 and 1 in 150,000 people worldwide<sup>1,2</sup> Mortality resulting from laryngeal edema and asphyxiation occurs in approximately 30% of affected individuals.<sup>3</sup>

A serious issue for individuals with HAE is a delay in diagnosis, taking an average of 8 years from the first hospital visit<sup>4</sup>. This delay is caused by several factors: HAE can resemble other conditions, such as allergies or stomach problems; symptoms are widely variable; and HAE is a rare disease and therefore not well known among physicians.

In 2008, a nationwide survey on HAE was administered to Japanese physicians of 4,495 responding physicians, 55.2% reported no knowledge of HAE<sup>5</sup>. To improve

diagnostic time frames, HAE-related educational activities for physicians commenced in 2008 with the support of a pharmaceutical company (CSL Behring K.K.). Nationwide seminars on HAE were held at physician meetings and at National Scientific Society conferences. Lectures on HAE were also held for the mass media and articles on HAE were published in journals and books. Educational brochures and posters on HAE were distributed to physicians at scientific meetings and by mail. A website entitled "HAE Information Center" was established in 2011 with website visits reaching 8,200 per month by 2014 (**Figure 1A**). A sudden increase in the number of website visit was noted twice. The first increase occurred when an explanation of Quinke's edema, another name for HAE, was added to the website (June 2013) and the second increase coincided with website access becoming available by smart phone (May 2014). Guidelines for HAE were established by the Japanese Association for Complement Research and published in 2012.<sup>6</sup>

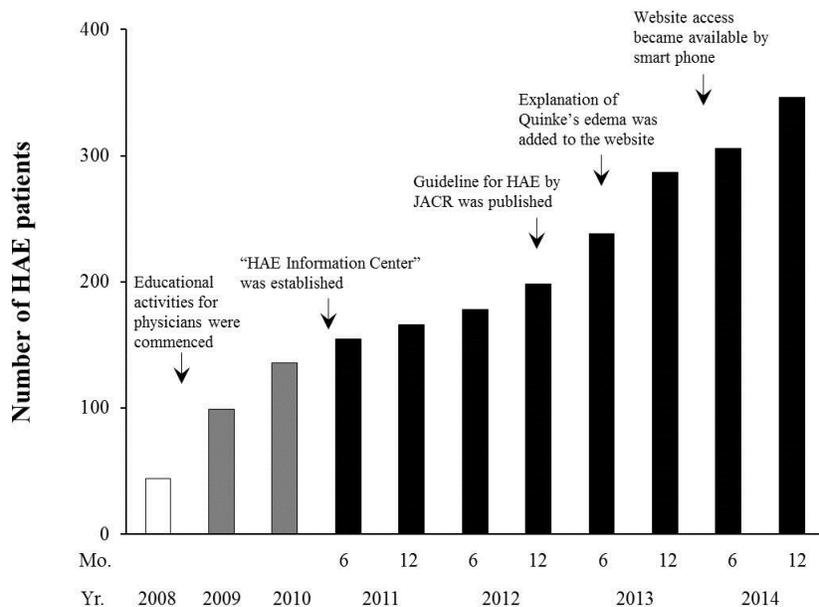


**Figure 1A.** Since the establishment of the HAE Information Center in 2011, the number of visits has increased.

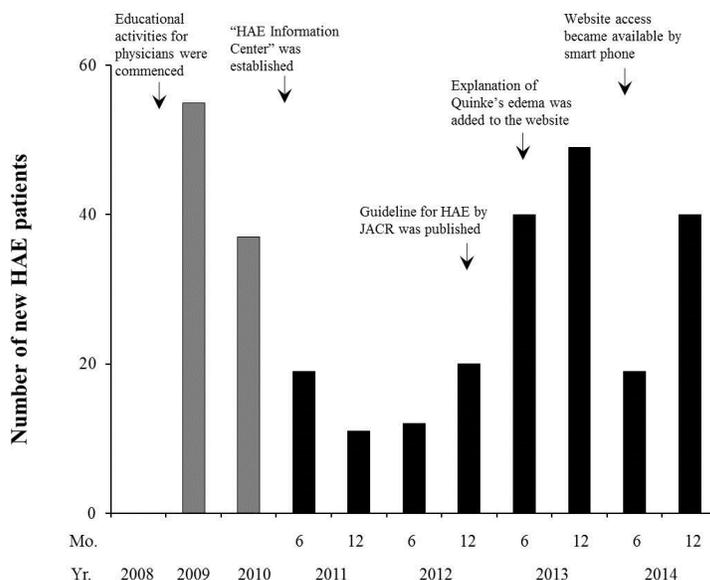


The educational activities listed resulted in an eight-fold increase in patients diagnosed with HAE in Japan (Figure 1B). There were no new HAE cases diagnosed in 2008 before the educational activities were initiated (Figure 1C), and the number of new patients diagnosed with HAE was increased in 2009 and 2010. This increase may result from the HAE educational programs. The

second increase in the diagnosis of new HAE patients was seen after the HAE website was established (2011). This study suggests the effectiveness of educational activities in assisting in diagnosis of rare conditions, and similar programs should be initiated for other rare disease to reduce misdiagnosis and thereby improve patient care.



**Figure 1B.** The total number of HAE patients has increased consistently since the commencement of the HAE educational program. The open bar shows the number of HAE patients before commencement of educational activities, grey solid bars show numbers of new patients after commencement, and black solid bars show numbers after website launch.



**Figure 1C.** There were no new patients diagnosed with HAE in 2008 before the educational activities commenced. After initiation of educational activities the number of new patients was increased. The second increase in new HAE diagnoses occurred after the website was established. Grey bars show numbers after initiation of educational activities, and solid bars show numbers after establishment of the website.



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