Embargoed until 12.01am Monday May 16, 2022

Access to affordable treatment critical for rare swelling disease.

HAE Day, May 16, 2022, for Hereditary Angioedema Awareness

“There is a huge difference between just surviving and properly living – and access to critical treatment is the difference between the two,” says Jess Bogoyevitch, a 22-year-old with hereditary angioedema (HAE), a very rare and potentially life-threatening genetic condition that leads to painful swelling in various body parts. Airway swelling is particularly dangerous and can lead to death by asphyxiation.

“I was having daily swelling attacks in my stomach, hands, feet and sometimes my whole body. The swelling could last for days, causing excruciating pain and gut issues. It can be very scary living in constant fear of an attack occurring,” explains Jess. “The attacks would also make me extremely sluggish. I was so sick that it was difficult to get out of bed for days at a time. It is really hard for people to comprehend the impact of living with HAE, where one day you can’t get out of bed and the next day you could be good again. Being such a rare condition, I have to do a lot of self-advocacy with paramedics and doctors – they just don’t know about it.”

Hereditary angioedema occurs in about 1 in 10,000 to 1 in 50,000 people. Symptoms include episodes of swelling in various body parts including the hands, feet, face and airway. Patients often have bouts of excruciating abdominal pain, nausea and vomiting that is caused by swelling in the intestinal wall.

Jess had a breakthrough five years ago when a new treatment drastically improved her quality of life. “Having access to the right treatment is life-changing. I had tried a few different treatments over the years as they became available, but they tended not to last for long or were difficult to administer. Finally, I had access to one treatment that really made a huge difference and I have never been more stable. I rarely have a major attack anymore.”

Fiona Wardman, CEO of HAE Australasia, lives with HAE herself and says the disease is poorly understood and often misdiagnosed.

“After many years of unexplained swelling attacks, I finally received a diagnosis 26 years after my first attack. This is not uncommon, with the average time to diagnosis historically being 13 to 21 years[i],” says Ms Wardman. “Internal swellings are not very obvious and may be diagnosed as psychosomatic, resulting in referral for psychiatric evaluation, or misdiagnosed as other illnesses, such as irritable bowel syndrome or allergies. Some people go through the unnecessary trauma of exploratory surgery because abdominal HAE attacks mimic a surgical abdomen, yet a simple blood test will determine if a patient has HAE.”

The attack rate varies from family to family, but some people can have several attacks per month making it very difficult to sustain a job and live a normal life. For people with frequent attacks, preventive therapy can be administered on a short-term or long-term basis, but access and affordability present great challenges.

Experts advocate for equitable access to treatment

Hereditary Angioedema Day or "HAE Day", on May 16, focuses on raising awareness about this rare disease. This year, experts are calling for equitable access to life-changing treatment.

Professor Connie Katelaris, HAE expert and Chair of the HAE Working Party for the Australasian Society of Clinical Immunology and Allergy, says "All Australian patients should have access to state-of-the-art treatment. We now have great understanding of the biochemical pathways that lead to the swelling and a number of drugs have been developed to block key molecules in the pathway. One of these is remarkably effective at blocking swellings and is now available in many countries for prophylaxis. For those who have frequent attacks, this drug will be a life changer enabling them to lead normal lives. While the drug was registered by the Therapeutic Goods Administration at the beginning of 2019, it took almost two years for it to be PBS listed at the end of 2021."

"This is one example of why we desperately need a more efficient approach for drugs for rare diseases. We need to limit the delay for all Australian patients to access state of the art treatment. Economic arguments alone are not appropriate for rare disease drugs," continued Professor Katelaris.

Equitable access to novel therapies such as gene therapies also needs to be considered.

"Gene therapy is being developed for a number of rare diseases including HAE and other primary immunodeficiencies. When the trials have been completed, we will need processes in place to allow equitable access to what will be curative therapy," continued Prof. Katelaris. "Clinical trial data is exceptionally difficult to obtain for rare diseases so we also need to set up structures to accept things such as genetic analysis and target molecule identification as an evidence base. Funding basic research for these rare diseases continues to be a priority as the benefits and knowledge gained flow to other areas of medicine as well."

Ms Wardman agrees, adding "Treatment is improving all the time, but equitable access is critical – it can save lives. As an example, before therapy became available, the mortality rate for airway obstruction was reportedly as high as 30 percent. Preventing or minimising attacks can also help people actually participate in life – it can enable people to do things that many of us take for granted, such as having a job, going to university or having a family."

Access to improved treatment for primary immunodeficiencies, including HAE, is a key focus of the new ASCIA Immunodeficiency Strategy, developed in collaboration HAE Australasia, and is available at www.nationalimmunodeficiencystrategy.org.au

Further information on HAE is available at https://haeaustralasia.org.au

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Distributed by Lanham Media on behalf of the Australasian Society of Clinical Immunology (ASCIA).

Media contacts:
Greg Townley | greg@lanhammedia.com.au | 0414 195 908
Fleur Townley | fleur@lanhammedia.com.au | 0405 278 758

NOTES FOR MEDIA:

Available for interview:

- Prof. Connie Katelaris AM (NSW) – Clinical Immunologist, HAE expert and Chair of HAE Working Party for the Australasian Society of Clinical Immunology and Allergy (ASCIA). Past ASCIA President.
- Fiona Wardman, CEO of HAE Australasia, lives with HAE herself.
- Case study: Jess Bogoyevitch, 22 years from Sunnybank/Brisbane, QLD

Additional case studies:

- Maureen, 73-year-old woman from South Albury NSW that was diagnosed when she was 16 and has experienced many life-threatening attacks that required trips to the hospital in ambulances and helicopters. She would frequently need to stay in bed for 5-10 days. Sometimes her throat would close – this happened three times in one year. Medical technology has improved substantially. She still gets attacks around once a month, but they are less severe thanks to treatment. She was the first in her family to be diagnosed. Her father, aunt and grandfather were subsequently diagnosed.
- Larissa (33, Perth WA). She had her first attack at age 11 or 12. She remembers being at school and felt her skirt getting tighter. She started vomiting and had waves of pain. She lived in a small town and had to get an ambulance an hour out of town. Since then, she has had quite violent stomach attacks and almost always ended up in hospital. Larissa's Mum and sister also live with HAE. Her family had the mind frame of just dealing with it on their own when they could because it was easier - the medical profession rarely understood. Treatment has improved her quality of life, but she had a life-threatening attack just last year – she thought she was going to die because no one in the hospital realised how critical it was for her to get the treatment she needed.
- Emily (23, Perth WA). Read Emily's story available here: https://haeaustralasia.org.au/emilys-story/

The Australasian Society of Clinical Immunology and Allergy (ASCIA)

The Australasian Society of Clinical Immunology and Allergy (ASCIA) is the peak professional body of clinical immunology and allergy in Australia and New Zealand. Established in 1990, ASCIA is a world leading, innovative and active professional society with strong leadership and sustainable operations.

ASCIA's purpose is to advance the science and practice of allergy and clinical immunology, by promoting the highest standard of medical practice, training, education and research, to improve the health and quality of life of people with immune system disorders, including allergy and primary immunodeficiency (PID).

ASCIA is committed to providing high quality training, education and research to improve the health and wellbeing of all people in Australia and New Zealand with PID.

Further background:

- Estimates of the occurrence of HAE range from 1 in 10,000 to 1 in 150,000 people with approximately the same frequency in men and women. Based on the current quoted number of cases in other populations, there could be up to 480 cases in Australia and 90 in New Zealand.[i]
- HAE patients have a defect in the gene that controls a blood protein called C1 Inhibitor. The genetic defect results in production of either inadequate or non-functioning C1-Inhibitor protein, which would normally help to regulate the complex biochemical interactions of blood-based systems involved in disease fighting, inflammatory response and coagulation.
- HAE is called hereditary because the genetic defect is passed on in families. A child has a 50 percent possibility of inheriting this disease if one of the parents has it. The absence of family history does not rule out the HAE diagnosis, however. Scientists report that as many as 25 percent of HAE cases result from patients who had a spontaneous mutation of the C1-

Inhibitor gene at conception. Consequently, these patients can pass the defective gene to their offspring.

- HAE can be triggered by emotional or psychological stress; physical stress (for example: injuries, surgery, repetitive or manual work), viral or bacterial infections; ACE inhibitors for high blood pressure, or oral contraceptives (the Pill).
- More than 50% of patients experience first symptoms of HAE before age of 10. 35% of patients experience first episodes in their teenage years and 7% occur in the first year of life. Historically, the average time to diagnosis has been 13 to 21 years but with increasing awareness there has been improvement in this delay in diagnosis.**Error! Bookmark not defined.**

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[i] https://haeaustralasia.org.au/wp-content/uploads/2019/11/Draft-Flyer-Double-Parrallel-Fold-About-Hereditary-Angioedema-master.pdf