

# ESTABLISHING A HEREDITARY ANGIOEDEMA PATIENT REGISTRY



*Assisting research that will lead to an improved quality of life for patients*



HAE Australasia Ltd  
Pre-Budget Submission for the  
Australian Federal Budget 2016/2017

Prepared for the  
Australian Federal Government  
February 2016

## EXECUTIVE SUMMARY

Hereditary Angioedema (HAE) is a rare, life-threatening, genetic medical condition which causes debilitating swellings around the body, including the airway which is life-threatening. Symptoms are often incorrectly treated as allergies or abdominal complaints which result in inappropriate treatments or unnecessary medical investigations. Where patients have an undiagnosed family history, they, on average, suffer symptoms for up to two decades before being correctly diagnosed - in the meantime suffering horrific symptoms that incorrect treatments don't resolve and wondering when the symptoms may appear next.

With the aim of improving the quality of life for HAE patients, 5 volunteers came together and established HAE Australia Ltd, now known as HAE Australasia Ltd, in 2011. As a result of the various projects conducted by HAE Australasia, there has been a significant improvement in access to information and life-saving medication for HAE patients in Australia. However, Australia is still far behind world best standards of treatments and management of HAE.

We are requesting that the Australian Federal Government provide our organisation, **\$40,000** during the 2016/2017 financial year to help cover the costs of a HAE Patient Registry which will improve the quality of research and help to increase the amount of research completed on HAE.

As a result of the increased quantity and quality of HAE research, more treatment options will be found, better management of HAE can be determine and further understanding of the condition will be available - all of these outcomes can improve the quality of life for HAE patients. Together, HAE Australasia and the Australian Federal Government can see this happen.

### Recommendation: Provide Financial Support for a HAE Patient Registry

**Project:** Patient Registry

**Project Details:** A HAE specific Patient Registry to house HAE patient personal medical data that, where appropriate, can be made available for HAE research

**Relevant Current Government Policy:**

Department of Health Goal - Outcome 1 - Population Health

Department of Health Goal - Outcome 2 - Access to Pharmaceutical Services

Medical Research Future Fund

**Estimated Development & Implementation Cost (\$):** \$40,000 - \$100,000

**Estimated Annual Maintenance Costs (\$):** \$2,000 - \$5,000 p.a.

**Australian Federal Government Contribution (\$)** **\$40,000**

**Funding Required By** Quarter 1, 2016/2017

# TABLE OF CONTENTS

Executive Summary.....	2
What is Hereditary Angioedema? .....	4
HAE Australasia – Our short history .....	5
Recommendation –HAE Patient Registry .....	6
Where funds will be sourced after the end of the proposed Government support:.....	8
Current Government Policies Affected By This Recommendation .....	8
Conclusion... ..	11
Appendix 1: HAE & The Patient Experience.....	12
Appendix 2: Team of HAE Australasia .....	14
Appendix 3: HAE Australasia’s Mission, Vision, Objectives .....	16
Appendix 4: HAE Australasia Projects & Outcomes .....	17
Appendix 5: Sample of responses from Research on the Australian HAE Treatment Landscape (2) .....	21
Appendix 6: Testimonials.....	23
References.....	30

## WHAT IS HEREDITARY ANGIOEDEMA?

Hereditary Angioedema (HAE) is a rare, genetic, medical condition where patients have a deficiency or abnormal function of the protein C1 Inhibitor. This condition causes spontaneous and recurrent episodes of swelling of various parts of the body, including:

- Airway, tongue or face – these swellings, if not treated quickly or appropriately, may lead to the patient dying due to asphyxiation.
- Intestinal swelling - causes immense pain, vomiting, diarrhoea and dehydration
- Other areas of the body (hands, arms, shoulders, torso, genitals, legs, feet) - even though swelling in these areas may not be life threatening, they may prevent the patient from undertaking every day activities including working or caring for their family.

For patients without a HAE diagnosis, the above symptoms may be treated incorrectly as allergies, abdominal complaints or injuries - treatments for these conditions to not relieve the HAE patient of their suffering. Unnecessary medical investigations may also be performed if HAE is not considered.

Studies have revealed that approximately 50 – 75% of HAE patients suffer at least one life threatening attack at some point in their life (1). Some patients suffer airway attacks on a more frequent basis (weekly, monthly or yearly) which leads to a serious fear of what the future may hold for them and if treatment will be available in time to save their life.

HAE attacks can be recurrent and spontaneous – there may or may not be a trigger for a swelling to occur, therefore, it is very difficult to predict when a HAE attack may occur. Life for HAE patients can have many unexpected and debilitating interruptions.



HAE is a rare condition – it may only affect approximately 480 people in Australia (1). However, these 480 people still deserve the same quality of life as those who were genetically given good health. Further information about patient experiences with HAE and how it affects their quality of life can be found in Appendix 1: HAE & The Patient Experience.

## HAE AUSTRALASIA – OUR SHORT HISTORY

Up until recently, the situation for Australian HAE patients was critical - they were faced with health care providers that had little to no knowledge of the condition, diagnosis was delayed and therefore patients endured years of suffering and treatment options were very limited.

However, in 2011, a group of 5 volunteers formed HAE Australia Ltd. Our vision was to enhance the quality of life for those living with HAE in Australia. Information about the Directors of HAE can be found in Appendix 2: Team of HAE Australasia



By 2013, we saw that the need of the 90 estimated New Zealand HAE patients was as urgent, if not more so, than Australia's and decided to include New Zealand in our activities. This led to a change of our organisations name – we are now known as HAE Australasia Ltd.

### ***Our Mission:***

*To help Australian and New Zealand patients gain subsidised access to more treatment options, empowering patients through education about their condition, educating the medical and general community about HAE to ensure patients are recognised and diagnosed more quickly, and to connect patients and carers with our support network – they need to know that they are not alone.*

See Appendix 3: HAE Australasia's Mission, Vision, Objectives further details

## HAE Australasia's Achievements So Far

Prior to HAE Australasia, there was NO representative body in existence in Australia and New Zealand for HAE. HAE patients had almost no contact with anyone else with the condition and the information that they had may have been provided by a health care provider with little to no knowledge themselves of HAE. Patients were not receiving adequate care.

HAE Australasia is governed by a board of 5 women who are either HAE patients or carers. We all volunteer our time outside of work and family commitments and are very passionate about making a difference. This is evidenced by the phenomenal achievements that have been made in the short time since our organisation was formed which include establishing a support network, developing a dedicated HAE website, playing a pivotal role in seeing new treatments becoming accessible to patients and assisting with HAE research (see Appendix 4: HAE Australasia Projects & Outcomes.

Despite these successes, there is still a long way to go to see that HAE patients' quality of life is improved and the standard of Australia's management of HAE is substantially enhanced to meet world best standards. With a relatively modest investment of funds from the Australian Federal Government, we at HAE Australasia believe we have the skills and networks to achieve significant improvements over the present situation.

## RECOMMENDATION –HAE PATIENT REGISTRY

HAE Australasia believes that a crucial element in improving the quality of life for HAE patients is to increase the amount of research that is undertaken about HAE and to ensure that research is conducted using a larger pool of quality information. This can be achieved by implementing a HAE Patient Registry that can store information about patient experiences and be provided to researchers to enable their finite resources to be used more effectively.

Prior to the inception of HAE Australasia, the handful of medical professionals and researchers that wanted to conduct HAE investigations in Australia had a limited patient base to contact. They may have only had contact with HAE patients known to them, or accessed patients via a Primary Immunodeficiency register. This database had 66 HAE patients, out of the potential 480 HAE patients in Australia (1) .

HAE Australasia is working towards being in contact with most, if not all HAE patients in Australia to provide a support network and education about the condition. Since our inception in 2011, our database has grown to over 160 patients and carers - including an Australian patient database of over 100 patients - this number already surpasses that of the Primary Immunodeficiency register.

By having access to a large and growing number of patients, HAE Australasia is in the best position to implement and maintain a Patient Registry with a substantial quantity of patient data available for HAE research which can lead to improved treatment, management and education of HAE (and therefore improving patient quality of life).

Australian Federal Government funding towards this recommended project will ultimately lead to patients being healthier, more active and have an increased participation in the community and economy. Future generations of HAE patients will also be in a far better position than those that have endured this condition so far.

*“One can only imagine the progress this energetic and capable group could make in key areas such as patient identification, education, diagnosis, and research if they had access to more funding sources.”*

*“It is remarkable that this intrepid group of dedicated volunteers has made such great strides in transforming the lives of a very sick and neglected patient population with relatively scarce financial resources.”*

## Project - Patient Registry - Development & Implementation

### *Details of Project:*

A HAE specific Patient Registry to house HAE patient personal medical data that, where appropriate, can be made available for HAE research. The Registry will be subject to regular upkeep and maintenance to ensure information is complete and readily available for HAE research. The Registry will comply with applicable ethical, medical, privacy, regulatory and other codes, guidelines and requirements.

### *Purpose of Funding:*

To ensure that the HAE Australasia Patient Registry can be developed to the highest standards. Knowledge gained from the development of this registry can be shared with other similar organisations to help them achieve the high standards expected of this project.

### *Budget for Project:*

Estimated expense: Development & Implementation:	\$40,000 - \$100,000
Estimated expense: Annual Maintenance Costs	\$2,000 - \$5,000 p.a

<b>Total amount requested from the Australian Federal Government</b>	<b>\$40,000</b>
Amount that will be secured from other sources by HAE Australasia:	Balance of Costs

### *Funding Required By:*

Quarter 1, 2016/2017

### *Government Legislation Changes Required?*

No - HAE Australasia's activities simply need to be considered as part of budgets

### *Measuring The Project's Success:*

Factors that will help us determine how successful this project is included (but not limited to):

- Ease of use for patient data entry (including time taken and clarity of questions/answers)
- Accuracy rate of data input
- Ease of use for reporting details at varying levels of authority (including time taken and clarity of questions/answers)
- Relevance, reliability and accuracy rate of filtered/reported information
- User feedback on data input process, data update process, data extraction process
- Uptake by current patients and carers on our database
- Continued use of the registry by patients and carers
- Uptake by researchers to use the resource
- Number of researchers that utilise the registry for more than one project
- Awareness of patients, carers, health care providers and researchers of Patient Registry's existence.
- quantity of different reports available

HAE Australasia is strongly focussed on monitoring progress and improving processes wherever possible. The organisation regularly assesses project outcomes, effectiveness and efficiencies – this project would simply be included in the monitoring process.

## Where funds will be sourced after the end of the proposed Government support:

Building secure sources of funding for HAE Australasia is currently underway. This includes strengthening relationships with grant providers and regular donors. It is anticipated that, once the registry is established with the requested Government support, our organisation would be in a position to fully finance the ongoing maintenance of the Patient Registry from other sources.

## Current Government Policies Affected By This Recommendation

### *Department of Health Goals - Outcome 1 - Population health*

Due to the uncertainty of HAE, patients often lead a very sedentary and unhealthy lifestyle and may be unable to participate in the work force due to the unpredictable nature of their condition.

This sedentary lifestyle increases the patient's risk of other avoidable illnesses and chronic diseases that are listed as "National Health Priority Areas" including:

- Mental Health
  - o HAE patients are known to suffer anxieties and depression due to the affect that their condition has on their life
- Obesity
  - o Reducing physical activity to avoid triggers of HAE attacks increases the likelihood of becoming obese. Obesity may then also lead onto the following illnesses.
- Diabetes
- Cardiovascular Health Issues
- Cancer

By educating patients, carers and health care providers about the condition, treatments required, ways that patients can cope with the emotional stress of having a life-threatening condition, as well as providing comprehensive education about triggers of HAE attacks, patients will have the tools and support to lead a more active and healthy lifestyle.

These positive effects would decrease the incidence of the other avoidable illnesses and chronic diseases listed. HAE patients could also increase their participation in the community and workforce as their symptoms may be more stable. This would lead to an increase in economic activity.

## *Medical Research Future Fund*

The preliminary/planning stage of the HAE Australasia Patient Registry is well underway and it is expected that the Patient Registry will be developed and implemented by December 2016.

We are proposing that, as part of the Medical Research Future Fund, the Australian Government allows support for the development/implementation of the Patient Registry. This support will help promote critical research required into HAE.

The Patient Registry will be developed after consultation with HAE experts, the Australasian Society of Clinical Immunology and Allergy, various universities and other research bodies, rare disease and genetic support organisations, other organisations that have successfully implemented Patient Registries and legal practitioners.

These discussions will help to ensure that the HAE Australasia Patient Registry is a world class resource that is compliant with all relevant privacy, ethical and medical codes of conduct/legislation.

HAE Australasia is in support of the Australian Federal Government's investment into medical research. However, as discussed in the McKell Institute's paper (3), even if research leads to new treatment options, it is of no use if the treatments aren't financially accessible to patients.

*"[HAE] research will be greatly facilitated by HAE Australasia in terms of reaching people who might be interested in participating in research, in particular if a register were established this would be extremely effective for that purpose".*

*"The HAE community is diverse and dispersed and individuals can be hard to reach but a register would facilitate contact and communication."*

## *Department of Health Goal - Outcome 2 - Access to Pharmaceutical Services - Life Saving Drug Program*

A HAE Patient Registry will enable researchers to have timely access to up to date and reliable HAE patient data. By taking away the lengthy process of trying to connect with HAE patients, researchers will be provided with more time, and better use of their resources, to further our knowledge of the condition. This will help in determining and developing new treatment options for patients.

As treatment options are limited at the moment, patients are having to endure some serious side effects in order to reduce their HAE symptoms. Having a broader range of treatment options will allow patients to find a treatment that best suits them and doesn't lead to further issues (that may increase dependency on the health system for other reason and not HAE)

*"When I was 24, I had to have half of my liver removed due to the side effects of the only long-term medication available that actually helped my HAE. I was in hospital for a month and then off work for another two months. Then, when I went back to work, I would be sick a few days every fortnight because we couldn't find another treatment that worked."*

*"I'm worried that when I get pregnant, I'll be so sick that I will be house bound and bed-ridden for the whole 9 months because I can't use any of the few medications that are currently available to me".*

*"I remember whenever my grandfather would have an airway attack, he would put a garden hose down his throat to make sure he could keep breathing - during those times, we didn't know where to go to get treatment or what the treatment was"*

*"I don't make plans with friends or family, just in case I'm sick again and I end up disappointing them - again"*

*"I don't travel anywhere that is more than an hour from a hospital that I KNOW has C1 Inhibitor - just in case I need it"*

## CONCLUSION...

Hereditary Angioedema (HAE) is a rare, life-threatening, genetic medical condition which causes debilitating swellings around the body, including the airway which is life-threatening.

This document discusses a recommendation of how the Australian Federal Government can support HAE Australasia to implement a HAE Patient Registry which can assist vital research by pooling HAE patient data into one resource. Research into HAE can help to improving the state of management of HAE in Australia and the quality of life for patients.

After reviewing our resources, it is estimated that a provision of only \$40,000 will help our organisation conduct this project

Together, HAE Australasia and the Australian Federal Government can further much needed HAE research, which can help Australian patients see an improved quality of life and enable an increased participation in their community.

## APPENDIX 1: HAE & THE PATIENT EXPERIENCE

### *HAE swelling attacks are usually spontaneous*

Even though the cause of HAE is known, what triggers a swelling attack varies from patient to patient and also, within a patient's own experience. What may have caused an attack at one time may not the next time. Often there is no apparent trigger.

The symptoms are often mistaken for allergies or abdominal complaints and are therefore, in the first instance, treated with inappropriate medical investigations or medications that either amplify the symptoms or are not effective and the patient continues to suffer. Patients MUST receive specific HAE treatments to reduce or prevent their symptoms.

### *Having HAE can severely affect a person's way of life (2)*

Where there is a family history of the condition, patients can be diagnosed early in life and can work with their doctor to put a medical action plan in place. It is often the case that the family doctor's only HAE experience is from other family members and may not be up to date with HAE best practice.

For the many where the condition hasn't been diagnosed, the patient may suffer decades of unexplained and debilitating swelling episodes that were either not treated correctly or resulted in unnecessary investigative procedures.

### ***Delay in Diagnosis***

On average, it takes 13 to 21 years (1) from the time a patient first experiences symptoms to the time a diagnosis is made. Even when a diagnosis is made, patients often encounter health care providers that have limited or no knowledge of the condition and therefore do not treat the patients with HAE specific medications. Significant improvements are required in this area to ensure that patients are not suffering or enduring unnecessary medical investigations which may only worsen their symptoms.

*“Doctors being unable to provide much support due to lack of awareness of HAE or expertise”*

*“[Patients have had] distressing experience at A & E. ... Lengthy delays, misdiagnosis, wrong treatment, anxiety”*

## ***Living with the threat of life-threatening or major attacks***

Once a diagnosis is made, patients live with the threat that a life-threatening or major HAE attack is about to occur. This results in severe anxieties and other mental health issues as well as a low participation in activities. Everyday occurrences that the general population take for granted can be an issue for HAE patients.

*"[There is a] negative impact on mental state .... Causing feelings of anxiety, depression, mood swings, inability to cope, relationship issues"*

*"Inability to plan in life; due to the degree of uncertainty and the unknown about attacks ... not joining clubs or classes, not planning longer term for activities or events, not taking up training or education"*

*"Periods of childhood or adolescence severely impact[ed] .... teasing, missing school, not being able to join in teams, not attending social events, minimising suffering and bravado. Detrimental impact on education"*

*"Restriction on [the] ability to travel to regional areas or overseas"*

*"Career limitation .... Not being able to work, taking a lesser career path"*

*"A curtailment of social life and an inability to participate fully in outside interest[s] .... Not being able to form friendships or attend functions"*

## APPENDIX 2: TEAM OF HAE AUSTRALASIA

Our team of 5 directors tirelessly volunteer time outside of family, work and other commitments. They co-ordinate all projects of HAE Australasia. Due to our own previous experience and enthusiasm to improve HAE patients' quality of life, we are all committed to adhering to best practice standards. We are very proud of the strong team that we have representing HAE in Australia and New Zealand. Our dedication is reflected in our successes and in the feedback we have received from HAE patients, carers and health care provider (see Appendix 6: Testimonials).

### *President - Fiona Wardman (Director)*

*Qualifications / Experience:* Office & Operations Manager  
Over 15 years' experience in Office & Project Management

Fiona's HAE diagnosis came in her 30's after years of unexplained swellings from the age of 9 which progressively worsened as she got older. No one else in Fiona's family has HAE. Fiona is proud to be part of an organisation that is making a difference to patients with HAE in Australia and New Zealand.



### *Vice President - Anne Wilkinson (Director)*

*Qualifications / Experience:* Qualified Pharmacist  
Over 30 years' experience in the field

Anne is married, has four adult children and lives in Melbourne. Anne was not diagnosed as having HAE until her late forties. Her diagnosis led to a diagnosis of other family members.



### *Treasurer - Louise Ridout (Director)*

*Qualifications / Experience:* CPA Accountant, Registered Tax Agent & Registered Financial Planner  
Over 10 years in public practice

Louise lives in Melbourne with her partner and young daughters. Louise was diagnosed with HAE when she was a baby as HAE was known in her family. She is passionate about raising awareness of HAE and keen to see better treatment options available for patients.



### *Secretary - Amanda Tionisio (Director)*

*Qualifications / Experience:* Office Manager for over 15 years

Amanda lives in Western Australia with her husband and 4 young children. She has a long family history of HAE and is particularly passionate about assisting children with HAE as she has two daughters with the condition.



### *NZ Representative - Olivia Willard (Director)*

*Qualifications / Experience:* Digital Marketing Specialist  
Over 12 years' experience

Olivia is based in Auckland, New Zealand and represents the interests of our New Zealand patients. She is passionate about New Zealand patients receiving and having access to the same treatment and care as patients in Australia and the rest of the world.



### *Medical Advisor - Professor Connie Katelaris*

Dr Katelaris is the Medical Advisor to HAE Australasia. She is Professor, Immunology & Allergy, University of Western Sydney, Head of Department and Senior Staff Specialist at Campbelltown Hospital. She is convenor of the Graduate Certificate in Allergic Diseases, the first postgraduate course in the Faculty of Medicine, University of Western Sydney. She is a past President of the Asian Pacific Association of Allergology, Asthma and Clinical Immunology, a board member of the Asthma Foundation NSW, and a past president of the Australasian Society of Clinical Immunology and Allergy.

# APPENDIX 3: HAE AUSTRALASIA'S MISSION, VISION, OBJECTIVES



HAE Australasia Ltd is a non-government, not for profit, health promotion charity that was established in 2011 as there was a need to increase the awareness of Hereditary Angioedema (HAE) in Australia and New Zealand.

## **Vision of HAE Australasia**

HAE Australasia Ltd.'s vision is to enhance the quality of life for those living with Hereditary Angioedema (HAE) in Australia & New Zealand.

## **Mission Statement**

To achieve this vision, HAE Australasia Ltd aims to help patients gain subsidised access to more treatment options, empowering patients through education about their condition, educating the medical and general community about HAE to ensure patients are recognised and diagnosed more quickly, and to connect patients and carers with our support network – they need to know that they are not alone.

## **Operational Objectives**

The objectives of HAE Australasia Ltd are to:

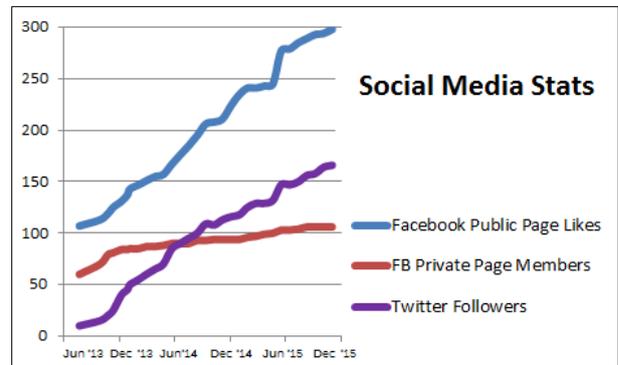
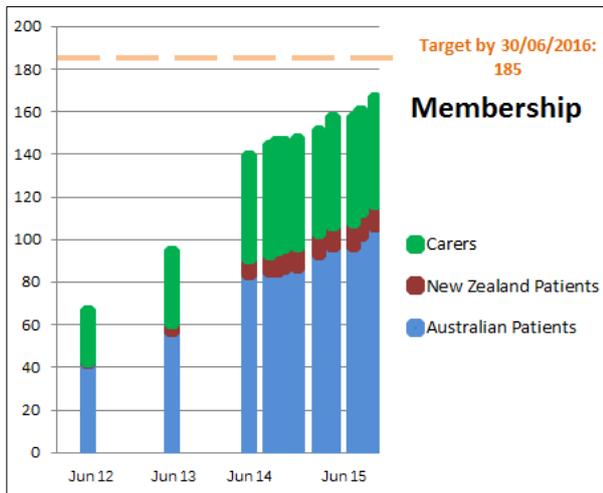
- Enhance the support and quality of life for people with HAE and their carers
- Provide a support group for people affected by HAE and allow patients and carers to communicate with each other to discuss their experiences. They need to know that they are not alone.
- Provide up to date and broad based education to:
  - o HAE patients to help them better understand their condition and treatment options available.
  - o HAE Carers to enable them to appropriately support individuals suffering from HAE
  - o Health care providers to enable them to appropriately recognise & support individuals suffering from HAE
- Engaging in activities to raise community awareness of HAE (to help undiagnosed patients access appropriate treatment and to help the community understand what it's like to live with the life-threatening and debilitating condition)
- Look at ways to make treatments and medications more accessible and subsidised for patients with HAE
- Assist research organisations with medical research into the causes, prevention and treatment of HAE
- Provide prompt and reliable responses in a professional manner to any HAE query
- Be considered by patients, carers, health care providers and the wider community to be a professional, reliable, courageous and compassionate organisation.

# APPENDIX 4: HAE AUSTRALASIA PROJECTS & OUTCOMES

Established an Australian & New Zealand support network of HAE patients & carers.

*"We support each other, because we know what it is like to have HAE."*

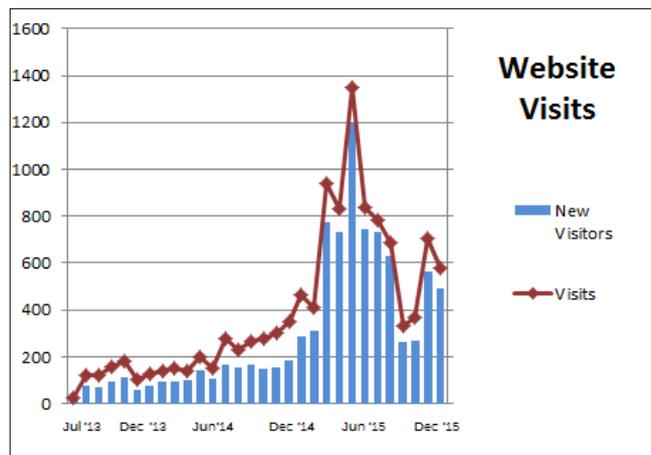
*"I don't feel so alone or isolated knowing there is an organisation and people in that organisation that understands and has knowledge about this awful disease that I live with every day."*



HAE Australasia Website - [www.haeaustralasia.org.au](http://www.haeaustralasia.org.au).

The aim is for this website to be the go-to place for all Australian and New Zealand HAE patients, carers, health care providers and supporters.

*"When I last saw my specialist for my check up, he used the HAE Australasia website to get resources that I needed."*



### *Assisted new treatments becoming subsidised*

We were actively involved and provided invaluable input into the applications of new treatments becoming subsidised. These treatments' becoming more accessible for patients was a major turning point for HAE patients in Australia and New Zealand - they felt that they could actually live a life. The following treatments are now subsidised (for specific uses):

- Firazyr (Icatibant) - Australia (2012)
- Firazyr (Icatibant) - New Zealand (2016)
- Berinert & Cinryze (C1 Inhibitor) - Australia (2016)

*"I can now afford to medicate myself and therefore have greatly enhanced my life. I no longer need to attend the emergency department of my local hospital, for treatment that lasts between a few hours to a couple of days, depending on the severity of the attack. I have done this at least once sometimes twice a month over the last 30 years or so. Which would of been of great cost to The Health System and the community."*

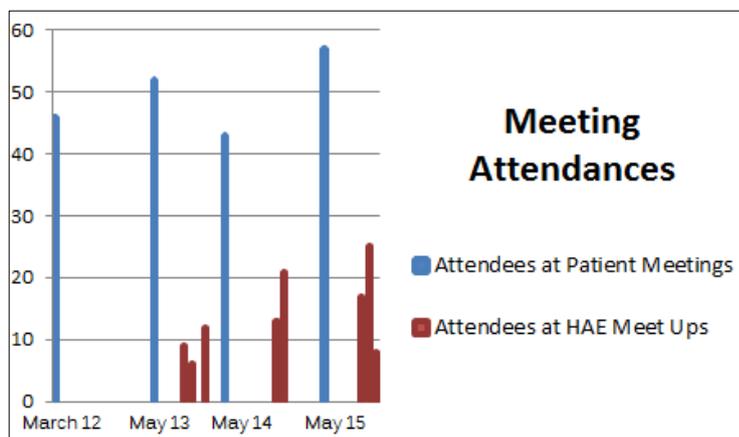
*"I have been involved in the introduction of new innovative therapies for HAE in Australia and negotiation with authorities can be difficult. Participation of an organized patient group facilitates such negotiation and can clarify the importance of new therapies to those with a poor*

*"Since the availability of Firazyr [Icatibant] for me which was October 2012 I have not attended the hospital for any medical treatment connected to my HAE. Thanks to the assistance of HAE Australasia."*

*"Having Firazyr available has lifted any anxieties I had about travelling away from a hospital that I knew had other emergency treatment. Thank you so much HAE Australasia for being my voice!"*

### *Held Patient Meetings & HAE Meet Ups*

Patient Meetings (conferences) educate patients and their carers about HAE, current research and new treatment availabilities. "HAE Meet Ups" strengthen the local HAE support networks.



Patient Meetings	Budget	Actual Spend*
2012 - Melbourne	\$9,600	\$8,407
2013 - Brisbane	\$14,360	\$13,490
2014 - Adelaide	\$26,200	\$14,270
2015 - Sydney	\$29,390	\$22,157

TABLE 1: BUDGET VS ACTUAL SPEND OF PATIENT MEETINGS. (\* HAE AUSTRALASIA WAS SUCCESSFUL IN APPLYING FOR OTHER GRANTS TO HELP FUND THESE PROJECTS)

*“Having been educated at HAE conferences and Forums I have attended over the years, for me knowledge is power and power is strength. Strength to carry on and live a happy, well and valuable life with this insidious disease.”*

*“Coming into contact with the HAE Australasia team has been life changing to say the least. They have given me the knowledge and the confidence to seek out the best medical treatment available”*

HAE Meet Up's*	Budget	Actual Spend
2013 - Perth, WA	\$600	\$555
2013 - Central Coast, NSW	\$200	\$131
2013 - Brisbane, QLD	\$1,000	\$927
2014 - New Zealand	\$2,260	\$2,043
2014 - Melbourne, VIC	\$1,900	\$1,580
2015 - Brisbane, QLD	\$1,900	\$1,339
2015 - Perth, WA	\$3,230	\$3,193
2015 - Central Coast, NSW	\$700	\$354

TABLE 2: BUDGET VS ACTUAL SPEND OF HAE MEET UPS (\* - HAE AUSTRALASIA HAS SECURED GRANTS TO HELP FUND THESE PROJECTS)

*“Whilst attending a HAE Australasia Patient meeting, I found out that the father of a toddler with HAE works in my building! What are the chances with so few HAE patients in Australia! It's good to know others are close by that I can talk to”*

*“Having HAE Australasia organising Patient Meet Ups has been invaluable for my self-esteem and feeling connected.”*

### *Increased awareness in the medical community*

There has been a significant increase in the number of doctors that now know about HAE due to our activities. This helps to identify potentially undiagnosed patients, or allows patients diagnosed with HAE be able to access appropriate treatments quicker.

*“There are now many more hospitals who are not only well educated about HAE but they also stock the correct medication.”*

### *Assisted with HAE Research*

HAE Australasia has connected research organisations with patients to conduct desperately needed HAE research and clinical trials. The quality of life and other issues surrounding HAE was discussed by patients in a recent research paper that our organisation helped locate participants for. Various responses to this research can be found in Appendix 5: Sample of responses from Research on the Australian HAE Treatment Landscape

### *Empowerment of Patients*

*“HAE Australasia is an important organization for the support and education of people with HAE and those associated with them, and for the furtherance of research and understanding of this condition”*

*“I really don’t know how I got on without HAE Australasia.”*

*“I would like to give my thanks and sincere appreciation to those involved with HAE AUSTRALASIA, as you have given me hope and an expectation for a better future.”*

*“I hold the HAE Australasia team in very high regard and I have absolutely no doubt that they saved my life, in more ways than one.”*

## APPENDIX 5: SAMPLE OF RESPONSES FROM RESEARCH ON THE AUSTRALIAN HAE TREATMENT LANDSCAPE (2)

- Patients reported that the top 3 sources of information were their specialist, online sources (including the HAE Australasia Facebook Page) and other family members
- There was a severe negative impact on the quality of life over the long term reported by HAE patients. Some of the issues that were identified included:
  - o “Periods of childhood or adolescence severely impact e.g. teasing, missing school, not being able to join in teams, not attending social events, minimising suffering and bravado. Detrimental impact on education”
  - o “Distressing experience at A & E. e.g. Lengthy delays, misdiagnosis, wrong treatment, anxiety”
  - o “Doctors being unable to provide much support due to lack of awareness of HAE or expertise”
  - o “Career limitation e.g. Not being able to work, taking a lesser career path
  - o “A severe impact on other family members. E.g. anxiety, impact on ability to work, disruption of childhood, necessity to care for grandchildren when a parent is ill”
  - o “The restriction on ability to travel to regional areas or overseas”
  - o “A curtailment of social life and an inability to participate fully in outside interest e.g. Not being able to form friendships or attend functions”
  - o “The inability to plan in life; due to the degree of uncertainty and the unknown about attacks e.g. not joining clubs or classes, not planning longer term for activities or events, not taking up training or education”
  - o “The decision not to have children (2 patients)”
  - o “Negative impact on mental state (not a discussion point but did emerge) e.g. Causing feelings of anxiety, depression, mood swings, inability to cope, relationship issues”
- “Stoicism is likely to have contributed to:
  - o Some limitations by some patients on their use of treatment e.g. because they have grown accustomed to dealing with attacks and one’s behaviour can be difficult to change.
  - o They may not feel an attack is severe enough in relation to other attacks they can have, even though most of the population might consider the same attack to be severe.
  - o They may not have discussed the true impact on life of their less severe attacks with specialists”
- Research found that, when determining the severity of attacks, not only were patients considering things like the impact on their airway, pain level, the location of the attack, the need to attend the emergency department, but they would also consider the impact that the attack had on their daily life, if they were able to continue looking after children or go to work. There were also instances where their specialist advice was in contradiction to the Australasian Society of Clinical Immunology and Allergy action plan and therefore wouldn’t treat an attack that may have been recommended by Australasian Society of Clinical Immunology and Allergy (or vice versa).

- “Health care providers were thought to be very supportive of self-administered treatment but some reported that their specialist was concerned about the costs and that they thought that this might be impacting them to discourage its “over use””
- The primary non product related barriers (to self-injecting Firazyr) were:
  - o “Perception of an attack not being severe enough”
  - o “Perception that the cost of Firazyr is high and so should be retained for the worst type of attack.”
- Secondary barriers:
  - o “Risk of running out of Firazyr for a really bad attack”
  - o ““Overuse” of Firazyr might lead to it’s being rationed or withdrawn”
  - o “Frequent use might reduce a patient’s pain threshold”
- The research found that, between specialists, nurses, other health care providers and patients, there was a discrepancy between how the severity of an attack was defined – this would impact when an attack was treated. For example:
  - o Location of the attack
  - o Mild v moderate v severe
  - o Mild v severe
  - o Non-significant v significant effect on quality of life
  - o Good v bad
  - o Making life “normal” for patient
- “Most Health Care Providers feel some pressure or responsibility to limit Firazyr recommendations”
- “Those (Health Care Providers) that recommend Firazyr only for airways/severe abdominal most cost conscious and more likely to talk about cost with patients”
- “Conversely, small number are not impacted by cost at all: These Health Care Providers think overall cost of treatment is low given low incidence of disease; cost savings given fewer hospital admissions and therapeutic benefit is high for patient cohort that has had inadequate treatment prior. Tends to be Health Care Providers that are more relaxed about patients using Firazyr to treat peripheral episodes”
- “Patients have learned to be stoic and to normalise symptoms”

## APPENDIX 6: TESTIMONIALS

### *1. Patient - Shane*

*HAE Australasia and me*

*January 2014 my daughter sent me an email saying she was going to apply for a travel grant to the HAE conference in Washington DC, and would I like to go too.*

*I looked at the application and found I was still in time to apply for a travel grant, and was able to get one. Without the grant, I would not have been able to attend. My daughter didn't attend due to other issues.*

*At the conference, I was sought out by the HAE Australasia people, as they knew I was attending, but I had not joined HAE Australasia. I would meet one person and then they said "you must meet this person". They were very passionate and made me feel special.*

*The conference was really an eye opener for me and I learnt so much.*

- *The president of HAEi (the international parent body), personally went around meeting everyone.*
- *Doctors gave me their contact details should I have issues (I went there on a fact finding mission to help my daughters' condition).*
- *I spoke to other patients from Australia and NZ...and around the world*

*Fiona [Wardman] told me I should be under an immunologist to manage my condition. I didn't fully understand this at the time as I have been on the same medication for over 30 years and had no issues. I followed her advice and discovered that one of the side effects of Danazol, is ischaemic heart disease and liver damage. I knew I had to get my liver functions tested, but never knew why. I now take a third of my original medication and have access to [Firazyr] in the event of an attack.*

*The information I have obtained, directly as a result of being a member of HAE Australasia is amazing. I have not only passed this onto my immediate family, but also other lines of my family.*

*My dad has seen a specialist and his medication has been reduced by half. He has recently had a triple bypass. I can't help thinking that maybe if we had made contact with HAE Australasia earlier and learnt more about HAE, then this may not have been necessary.*

*Now getting back to my fact finding mission for my daughter. She lives in Christchurch and up until last year, was a 2<sup>nd</sup> year law student. She has had her life turned upside down due to HAE. She is a member of HAE Australasia and has been supported by them. It wasn't until August last year that I realised how dire her situation was. She was depressed and had given up on life. I was so concerned that I flew to Christchurch from Melbourne to sort out her issues. Olivia Willard, HAE Australasia NZ Director, put me in touch with Dr Jordan, an immunologist in Auckland. I arranged for a doctors referral for my daughter to see him. Olivia arranged a patient meet-up for NZ, in Auckland. Dr Jordan organised an appointment a few days before the meet-up and HAE Australasia gave her a travel grant to fly up to Auckland. She stayed with my dad in Hamilton and they both travelled to the meet-up together. At the meet-up she met a young lady, also treated by Dr Jordan, and they compared notes.*

*It became apparent that my daughter's HAE management is not being managed well (at all).*

*At a recent visit to Christchurch hospital, she woke up in ICU with a target on her throat showing the location for an emergency tracheostomy. She was given the wrong medication, and not enough of the medication she needed. I am on my way to Christchurch to meet with the department heads at Christchurch hospital to sort this out.*

*Without the help, support and guidance that HAE Australasia has given my family, I would not be equipped with the information required to help my daughter and she probably would not be with us today.*

*I am able to suggest that the hospital emergency staff get some training on HAE, and offer the help and resources of HAE Australasia to do this.*

*HAE Australasia, via their face book page, gives me access to patient forums where I can compare notes with other patients. We support each other, because we know what it is like to have HAE.*

*Via the HAE Australasia website, there are a wealth of resources to help us deal with any situation.*

*We can get instant help via the forums or by emailing one of the team from HAE Australasia.*

*As you can imagine the worry I have about my daughters' condition, but HAE Australasia has been with me all the way with advice and support.*

*I really don't know how I got on without HAE Australasia.*

*Regards*

*Shane*

## *2. Patient - Mauree*

*To whom it may concern,*

*This TESTAMONIAL may be used, along with my name, for any reason by HAE AUSTRALASIA.*

*The following is a part of how my life has been improved by being a member of HAE AUSTRALASIA.*

*I don't feel so alone or isolated knowing there is an organisation and people in that organisation that understands and has knowledge about this awful disease that I live with every day.*

*The fact that HAE AUSTRALASIA has been involved in the process of getting my medication on the PBS has greatly improved my life.*

*I can now afford to medicate myself and therefore have greatly enhanced my life.*

*I no longer need to attend the emergency department of my local hospital, for treatment that lasts between a few hours to a couple of days, depending on the severity of the attack. I have done this at least once sometimes twice a month over the last 30 years or so. Which would of been of great cost to The Health System and the community.*

*Since the availability of FIRAZYR For me which was October 2012 I have not attended the hospital for any medical treatment connected to my HAE. Thanks to the assistance of HAE AUSTRALASIA.*

*Having been educated at HAE conferences and Forums I have attended over the years, for me knowledge is power and power is strength. Strength to carry on and live a happy, well and valuable life with this insidious disease.*

*Having HAE AUSTRALASIA Organising Patient Meet Ups has been invaluable for my self-esteem and feeling connected.*

*Being one of the senior members of the HAE Community, I have been able to pass onto the younger members some of the knowledge and awareness I have gained through these meetings.*

*For individuals we have no hope of consulting with government departments, health bodies and drug companies regarding research, development and funding for potential new treatments.*

*Without the Doctors, committee and supporters of HAE AUSTRALASIA who give their time willingly our lives would be very different.*

*I would like to give my thanks and sincere appreciation to those involved with HAE AUSTRALASIA, as you have given me hope and an expectation for a better future.*

*Yours sincerely,*

*Maureen*

### 3. Patient - Cindy

*Just a few years ago I was a patient in a very remote location living with a rare, life threatening disease, had no effective medication on hand, knew very little information about my disease and my health care providers didn't seem to be able to give me any answers either. I felt isolated, helpless and was a ticking time bomb waiting to go off. It was very much a case of "you don't know what you don't know". Coming into contact with the HAE Australasia team has been life changing to say the least. They have given me the knowledge and the confidence to seek out the best medical treatment available. I have learned that when you have a rare condition like HAE it is vital to be an expert on your own condition and not rely on health care providers to know what's best, as this is not always the case. The education of the patients has had a massive flow on effect to many medical professionals as well. There are now many more hospitals who are not only well educated about HAE but they also stock the correct medication. This is all thanks to the power of an organisation doing the talking, rather than coming from just one person. As a group they have been able to assist in the progress of new HAE medications being considered and also subsidised. The development of the website has made important information and documents such as the Action Plan more readily accessible. The online support group and yearly patient meet ups play an important role of making patients feel understood and that they are not alone, this in itself is a very powerful thing as many patients have never met another person with the same condition.*

*I hold the HAE Australasia team in very high regard and I have absolutely no doubt that they saved my life, in more ways than one.*

#### 4. Anonymous Patient Testimonials & Quotes

*“Having Firazyr available has lifted any anxieties I had about travelling away from a hospital that I knew had other emergency treatment. Thank you so much HAE Australasia for being my voice!”*

*“Pregnancy and breastfeeding a newborn is hard enough, let alone dealing with HAE and not being able to take regular medication to help it go away for a little while! If only C1 Inhibitor was subsidised so that I could get through this time in my life”*

*“It’s amazing that in countries as affluent as Australia & New Zealand, we are still so far behind the rest of the world with treatments for HAE”*

*“When I last saw my specialist for my check up, he used the HAE Australasia website to get resources that we needed for my HAE management plan.”*

*“I remember whenever my grandfather would have an airway attack, he would put a garden hose down his throat to make sure he could keep breathing - during those times, we didn’t know where to go to get treatment or what the treatment was”*

*“I don’t travel anywhere that is more than an hour from a hospital that I KNOW has C1 Inhibitor - just in case I need it”*

*“I’m worried that when I get pregnant, I’ll be so sick that I will be house bound and bed-ridden for the whole 9 months because I can’t use any of the few medications that are currently available to me”.*

*“When I was 24, I had to have half of my liver removed due to the side effects of the only long-term medication available that actually helped my HAE. I was in hospital for a month and then off work for another two months. Then, when I went back to work, I would be sick a few days every fortnight because we couldn’t find another treatment that worked.”*

*“Whilst attending a HAE Australasia Patient meeting, I found out that the father of a toddler with HAE works in my building! What are the chances with so few HAE patients in Australia! It’s good to know others are close by that I can talk to”*

5. HAEi (International) President / HAEA (USA) President - Anthony Castaldo,

*My name is Anthony Castaldo and I serve as the President of HAE International—the umbrella organization that represents the world’s HAE patient group. I am also the President of the United States HAE Association.*

*I have the great privilege of interacting with leaders and members of HAE patient organizations from throughout the world. I must say that the HAE Australasia group and their leadership stand out as one of the most well organized, energetic, and effective of all our member organizations. The progress HAE Australasia has made in serving the HAE patient community since its relatively recent inception is nothing short of extraordinary. With limited resources, this “can do” organization has made great strides in identifying patients and helping otherwise sick people receive desperately needed treatment. This is no small accomplishment because HAE is a rare, difficult to diagnose disease that is not only debilitating, but potentially fatal. It is remarkable that this intrepid group of dedicated volunteers has made such great strides in transforming the lives of a very sick and neglected patient population with relatively scarce financial resources. One can only imagine the progress this energetic and capable group could make in key areas such as patient identification, education, diagnosis, and research if they had access to more funding sources.*

*Sincerely.*

**Anthony J. Castaldo**

*President*



[www.haei.org](http://www.haei.org)

Email: [a.j.castaldo@haei.org](mailto:a.j.castaldo@haei.org)

Cell: +1 202 247 8619

6. Immunology Senior Consultant – Dr William Smith



**Government of South Australia**  
**SA Health**

29 January 2014

To Whom It May Concern:

RE: HAE Australasia

I am a medical immunologist working at the Royal Adelaide Hospital and in private practice. I am consulted by people who have HAE to provide medical management and medical advice.

I have found HAE Australasia to be an excellent organization, well organized and enthusiastically run. It has been very useful in providing high-quality information and support for people with HAE. The newsletters have been very effective in providing information on developments and events in the world of HAE. HAE Australasia seems to be well linked and integrated with equivalent international organisations.

I and my colleagues have from time to time carried out research with the purpose of improving management and conditions for people with HAE. This research will be greatly facilitated by HAE Australasia in terms of reaching people who might be interested in participating in research, in particular if a register were established this would be extremely effective for that purpose. The HAE community is diverse and dispersed and individuals can be hard to reach but a register would facilitate contact and communication.

I have been involved in the introduction of new innovative therapies for HAE in Australia and negotiation with authorities can be difficult. Participation of an organized patient group facilitates such negotiation and can clarify the importance of new therapies to those with a poor understanding of the condition.

In my opinion, HAE Australasia is an important organization for the support and education of people with HAE and those associated with them, and for the furtherance of research and understanding of this condition.

Yours sincerely

**Dr William Smith**  
**Immunology Senior Consultant**

**Adelaide Health Service**

North Terrace,  
Adelaide, SA 5000

Tel: +61 8 8222 4000  
Fax: +61 8 8222 5939

ABN 80 230 154 545  
[www.health.sa.gov.au/onahs](http://www.health.sa.gov.au/onahs)

**Internal Medicine Service**

**Outpatient Appointments**

Level 9 Outpatients

Tel: (08) 8222 5174  
Fax: (08) 8222 2433

**Immunology and Allergy**

**Secretary**

Ms W Sala

Tel: (08) 8222 2953  
Fax: (08) 8222 0020

**Head of Unit**

Assoc. Prof. Robert Hedde

MBBS FRACP FRCPA PhD

**Consultants**

Dr William Smith

MBBS FRACP FRCPA PhD

Dr Frank Kette

MBBS FRACP FRCPA PhD

Dr Pravin Hissaria

MBBS MD FRCPA FRACP

## REFERENCES

1. **Katellaris C, Smith W, Mullins R, Gillis D.** *Position Paper on Hereditary Angioedema.* s.l. : Australasian Society of Clinical Immunology and Allergy, August 2012.
2. **Perspective Research (commissioned by Shire Australia).** *Understanding the HAE Treatment Landscape & Impacts For Self Administration of Acute Attacks.* March 2014.
3. **McKell Institute.** *Funding Rare Disease Therapies in Australia.* November 2014.