



Patron Prof John Hopwood

Australian Pompe's Association Submission Issue 2

Pharmaceutical Benefits Advisory Committee (PBAC) Meeting

November 2010 PBAC Meeting



APA

AUSTRALIAN POMPE'S ASSOCIATION

Patron Prof John Hopwood

Dedication to Pompe Patients who have passed away since July 2008.



Chad was born in country NSW in 2008 with Infant Onset Pompe Disease and was diagnosed too late to get treatment and he lost his precious life at seven months.

CB Passed away in April 2010 at 57 years old.



JL passed away at age 47 in 2009 after many years of illness.

Our hearts go out to the families of these members who's loved ones have passed away since July 2008. These tragedies remind us all, how difficult it is to diagnose and treat such rare diseases as Pompe and how important it is to start treatment early. With Myozyme treatment we have the power to stop these tragedies occurring again and we must all endeavor to ensure that timely treatment is available to all Pompe patients.

Helen Walker OAM
President APA and IPA Board Member



Patron Prof John Hopwood

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Australian Pompe's Association Inc

Founded January 1998

Member of the International Pompe Association (IPA)

Registered Charity

| | |
|-----------------------------|-----------|
| Membership June 2010 | 22 |
| Infant onset | 2 |
| Juvenile Onset | 2 |
| Adult Onset | 18 |

| | | |
|-----------------------------|-----------|--|
| Members in treatment | 18 | |
| Infant Onset | 2 | Government Funded LSDA |
| Juvenile Onset | 2 | Genzyme International Charitable Access Program |
| Adult Onset | 14 | Genzyme International Charitable Access Program |

| | |
|-----------------------------------|----------|
| Members awaiting Treatment | 4 |
|-----------------------------------|----------|

| | |
|----------------------------------|----------|
| Members location by state | |
| New South Wales | 8 |
| Queensland | 6 |
| Victoria | 4 |
| Tasmania | 3 |
| South Australia | 0 |
| Western Australia | 1 |

Myozyme a Life Saving Drug Australia

In the last 18 months 2 Adult Onset Pompe patients being 10% of the APA membership have passed away due to Pompe Disease

The Pompe's community in Australia is small with 22 adult and juvenile onset APA known patients.

Both patients had received Myozyme treatment but unfortunately the delay of 9 years and 18 years from diagnosis to the start of treatment was too long for the treatment to halt the progress of the disease and major degradation and frailty had already developed.

Based on the UN life expectancy for Australia of 79 years for men and 84 years for women this is a reduction of 32% and 41% of the patient's predicted life span.



Patient one CB

Diagnosed with Pompe Disease in 1997 started Myozyme treatment 2006 and she passed away in 2010 age 57.



Patient two JL

Diagnosed with Pompe Disease in 1988, started Myozyme treatment 2006 and passed away in 2009 age 49.



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Myozyme a Life Saving Drug Sibling comparison

Patients receiving treatment with Myozyme live longer than siblings who did not have the benefits of treatment.

7 Cases of Adult/Juvenile Onset Pompe Disease patients who are currently receiving treatment and have a history of one or more siblings passing away due to Pompe disease have been identified.

These cases include

Case one RS Australia

Brother passed away never having Myozyme treatment at 54 years old. Patient RS has been receiving Myozyme treatment for three years, condition is stable with improvements in muscle and lung functions, age 59.

Case two TK United States of America

Sister passed away after only six months of Myozyme treatment at 39 years old. Patient TK has been receiving treatment for the last four years, age 40, condition has stabilised and lung function improved.

Case three CL Germany

Sister passed away at 33 years old after many years of critical illness and brother at 5 years old with symptoms from birth. Patient is ventilator dependant and has been receiving Myozyme treatment since 2006 now age 28. Except for some minor infections health has been good since being on treatment.

Case four MA United States of America

Sister passed away at 8 years old. Patient currently in treatment since 2003 now age 9 and progressing well.

Case five Netherlands

Brother passed away at 50 years old. Patient currently receiving Myozyme treatment and is now over 50 years old.

During our review of Siblings at no time have we identified any cases where a Pompe patient receiving treatment has passed away younger than a sibling who has been diagnosed with Pompe but was not receiving treatment.

This information was gathered with the assistance of the International Pompe Association and the various country organisations in the United Kingdom, Netherlands, France and United States of America.



Myozyme a Life Saving Drug Dangers of Frailty

Due to loss of muscle mass Pompe's patients who are not receiving Myozyme treatment are often frail and subject to infections, pneumonia and injury.

Details for 6 Australian Patients prior to Myozyme treatment are shown below.

| Patient | | Height | Weight | Australian Average Weight | Functional Vital Capacity (FVC) Actual | Predicted FVC | % |
|---------|--------|--------|--------|---------------------------|--|---------------|------|
| DW | Male | 185 | 52 | 84 | 1.97 | 5.26 | 37% |
| BD | Female | 180 | 63 | 68 | 3.77 | 4.66 | 80% |
| RS | Male | 177 | 67 | 84 | 2.44 | 4.3 | 57% |
| BC | Female | 168 | 86 | 68 | 0.81 | 2.81 | 29% |
| DD | Female | 162 | 49 | 68 | 2.32 | 3.66 | 63% |
| AR | Male | 181 | 72 | 84 | 5.32 | 5.34 | 100% |

AR shown above is a younger sibling of CB who passed this year, AR has not at this time been accepted for Myozyme treatment under ICAP.

ABS data

Between 1995 and 2004–05, the average weight (kg) of both male and female adults increased across all age groups. During this period, the average weight of an adult female increased from 65kg to 68kg, and for males it increased from 80kg to 84kg

FVC predicted National Asthma Council <http://www.nationalasthma.org.au/content/view/321/410/>

Myozyme a Life Saving Drug Treat Early

Through this information we are reminded of the 4 members of the Australian Pompe community diagnosed and who are still waiting to start treatment with Myozyme.

Every week these patients progress and become frailer, with Myozyme we can stop the progression and prolong and enhance the lives of these patients and their families.

Due to the delays in approval the Genzyme ICAP is now closed and there is no treatment available. This is a developing tragedy and we need to do all possible to obtain treatment for these patients. The Brad G story that follows is an example.



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**Mr Brad G Adult Onset Pompe Patient Buderim Queensland
Specialist Dr Jim McGill**

I was diagnosed with Pompe's disease at the end of May this year and I am currently not receiving Enzyme Replacement Therapy since new applications to Genzyme's ICAP are not being accepted at present.

I am currently still working full time as an electrician and providing for my family but I am struggling to do so because of the effects of Pompe's disease. The fear I have, is that with the rate of progression of my disease, I estimate I have 6 to 12 months before I will have to give up work due to escalating back pain and muscle weakness and that my condition will continue to deteriorate after this. My doctors tell me I have Obstructive Sleep Apnoea and the onset of Respiratory Failure and I need a Bipap machine to help treat this.

I am greatly encouraged by the degrees of improvement experienced by other Pompe patients receiving Myozyme. My hope is that I will be able to access Myozyme to halt my disease progression, improve my muscle strength and respiratory function. Hopefully, I will be able to keep working and have a long and fulfilling life with my young family.



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"Of the many challenging aspects of learning you're diagnosed with a neuromuscular disorder that is so rare, is understanding where to begin once the diagnosis is confirmed. It can be a major obstacle for many."

The Australian Pompe's Association helps with the valuable link between those newly diagnosed and getting the services and resources they desperately need.

The following people presented in this document have gone from the depths of despair to the optimism of hope of continuing treatment that has changed their lives.

These are the

TURNING POINTS
of our members



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**Mr Michael A Juvenile Onset Pompe Patient Townsville Queensland
Treating Centre Townsville Hospital Specialist Dr Lyndon Roberts**

Since I started treatment with Myozyme, I no longer have the need for a walking stick now and we have just purchased a home and plan to have more children and work for the all Australian dream of home and family.



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Diane D Adult onset Pome Patient Western Australia
Treating Centre Royal Perth Specialist Prof Phillipa Lamont

I was experiencing typical problems associated with weak respiratory muscles, waking several times a night with headaches and a heavy heartbeat, resulting in tiredness throughout the day. I had been prescribed a B-Pap machine but was having enormous problems adapting to the machine and breathing apparatus. After trialling many different models and styles my respiratory specialist concluded I may not be able to use a machine. This decision coincided with my commencing treatment with Myozyme in August 2009.

After only 3 treatments my breathing had noticeably improved and after 6-8 treatments I no longer woke with any symptoms, I am much more alert and do not experience tiredness during the day. I am far more energetic than I have been for a long time, I didn't realise how much I had lost until I experienced improvement.

I work fulltime in a management position with an International recruitment company. My work is demanding and requires me to work far beyond the statutory 40 hour week. I enjoy my work and if I can maintain my health I plan to work for several more years. I have two children, Amanda and Alister, we are a very close family and all live in Perth, Western Australia.



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**Michael C Adult onset Pompe Patient Tasmania
Treating Centre Royal Hobart Specialist Dr Dreyer**

I am 52year of age and have just completed my 2nd year of Myozyme treatment.

My personal experience has been a slow improvement in my physical and emotional condition I.E. Strength improving in my arms weight gain and my stability. I used to have frequent falls and injury but in the past year none.

My wife Leonie and I enjoy a independent life style on our 2 acre block growing our own fruit and vegetables self sustainably.

We had to retire from our agricultural and retail business eight years ago because of my health and my prognosis was bleak.

Now because of my treatment we have renewed confidence that we can continue to enjoy our independent lives.



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Raymond S Adult onset Pompe Patient NSW
Treating Centre Westmead Hospital Specialist Prof Silience

I was diagnosed with Pompe Disease in March 2002, I started treatment with Myozyme in February 2007 prior to this the progression of my disease had been very rapid and I was advised that my life expectancy was poor. Within 6 months of treatment the rapid progression of the Disease had stopped and I started to regain some of the strength I had lost.

Pompe Disease causes a patient to fade away in front of their loved ones. After many years of illness, my brother spent the last two years of his life in hospital under intensive treatment and intervention and passed away, at 54 years old, never having the benefit of Myozyme. I am 59 years old and every year that the Myozyme treatment gives me and my family, is a blessing.

I have a loving wife Christine and a 17 year old daughter who is currently studying for her HSC. Prior to the Myozyme treatment, I was concerned that I would not see my daughter graduate, now this milestone is possible.

I am very concerned that should Myozyme not be approved by the PBAC and Genzyme suspend my access to treatment under the ICAP program, my Pompe disease would rapidly return to its former progression and I would, once again, be facing a very uncertain future.

My experience is that Myozyme stops or delays the effects of Pompe and is a wonderful advance that should be available to all who need it



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Mr. Ari K Adult onset Patient Victoria

Treating Centre Monash Hospital Specialist Dr Andrew Churchyard

Before I started Myozyme, I had sought information on leaving my employment with Australian Post because of my illness.

I have improved so much that I am now able to continue working full time.

My family will benefit by this and my greatest wish is to see my 3 daughters educated to be guaranteed of good employment.



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Belinda D Adult onset Pompe Patient NSW
Treating Centre Westmead Hospital Specialist Prof Sillence

I started Myozyme after the birth of my daughter. My energy levels have increased and my feeling of well being is great.

I have confidence now, that I will see my children grow up, with my input of being a good Mum.



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First day of treatment 2nd August 2007



Conference call April 2010

Helen W Adult onset Pompe Patient Victoria
Treating Centre Frankston Hospital Specialist Prof E Butler

Myozyme changed my life from spending hours a day on the Bi pap to not needing it at all during the day.

I have been able to get back into the community, get out in the garden, go for a drive and the treatment allows me to undertake a conversation without losing my breath.

My future was so close to being put in a nursing home.

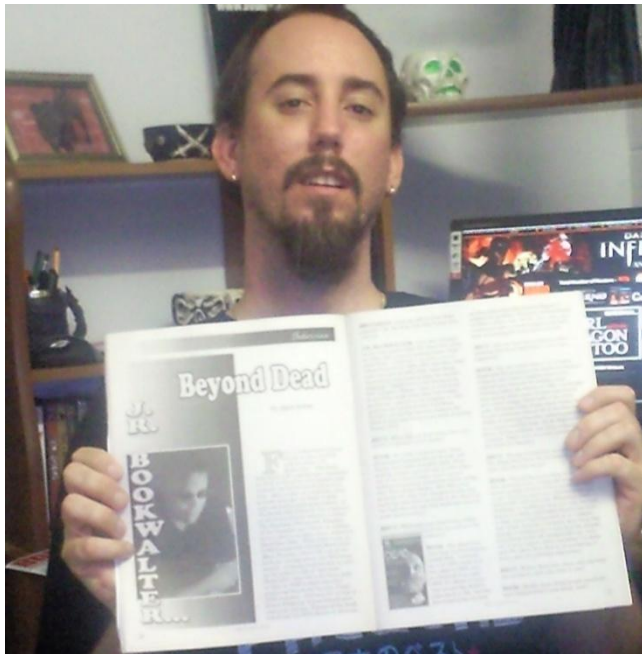
Yes a life saving experience, One that I will be forever grateful to Myozyme for.



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Jason A Juvenile onset Pompe Patient Queensland
Treating Centre Charters Towers Specialist Dr S Mallick

Since starting treatment I can now work 12 hours a day at my computer and I am establishing a web site programme to keep me busy.

I feel a different person now with so much more energy.



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Bet C Adult onset Pompe Patient New South Wales
Treating Centre John Hunter Hospital Specialist Prof Sillence

I have lived with Pompe Disease for a very long time and before Myozyme I was rapidly losing ground and going downhill fast. Most of the time, I was very tired, had no energy and was doing very little.

I started Myozyme infusions in 2006 and since then things have become much better.

My quality of life has much improved. I feel stronger in myself, have a lot of energy and feel just great. I am back doing some of the things I could no longer do and I can again enjoy spending time with my family and friends.

Life is good!



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Mr. Doug W Adult onset Pompe Patient Queensland
Treating Centre Wesley Hospital Specialist Dr Noel Saines

I was diagnosed with Pompe Disease when I was admitted to hospital after a cardiac arrest in December 08.

This was caused by severe breathing difficulties as a result of the damaging effect of Pompe on my diaphragm.

I began Myozyme treatment in March 09 and within 2 months was back to work on a part time basis which has now increased to full time.

My health and physical fitness is returning to their levels prior to the onset of myPompe disease.

Observations on a patient that is no longer able to receive ERT

- **Patient Age 47**
- **Started Treatment Feb 2005 in Australia**
- **At time of first treatment patient was mobile using and electrical wheelchair and was ventilated.**
- **Treated for 4 years and condition was stabile.**

In 2009 the patient, due to family situation, left Australia and was no longer able to have access to ERT

6 months after ceasing Myozyme treatment

- **Patent is confined to bed day and night.**
- **Suffers from bed sores, contractures, stiff muscles.**
- **Unable to move hands.**
- **Requires a PEG tube, sore suctioning area etc.**
- **Patient is unable to talk and requires frequent suctioning.**
- **Patient is now only 39 kilos and is fed via PEG protein rich fluids.**
- **Required hospitalisation twice to remove fluid from abdomen.**



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CobWebs, 43a North Lane, Buriton, Petersfield, Hampshire
GU31 5RS, United Kingdom. www.Pompe.org.uk



Luitenant Generaal van Heutszlaan 6, 3743 JN Baarn

Helen Walker, President
Australian Pompe Association
4/55 Van Ness Avenue, Mornington,
Victoria 3931, Australia

14th January 2010

Dear Helen,

Re: Approval of Myozyme, Enzyme Replacement Therapy for Pompe Disease

I am writing to you in my capacity as chairman of the International Pompe Association (IPA); a federation of support groups for families affected by Pompe Disease covering over forty countries around the world. I also support patients in the United Kingdom through my work for the Association for Glycogen Storage Disease (AGSD-UK).

I would like to confirm the International Pompe Association's support for the Australian Pompe's Association application for Myozyme to be included in the Australian Pharmaceutical benefits system. I am concerned and dismayed that a rich, developed country like Australia does not yet support its small number of Pompe's patients, while much poorer European countries are now doing so without complaint. With Australia's excellent history of Pompe's diagnostic research and world leading capability through Professor Hopwood's group at the Adelaide Children's Hospital I would have expected Australia to have been a leader in its support of its Pompe's Patients.

Through my work for the AGSD-UK and the IPA I continually hear reports from many patients of all ages, infants through to elderly pensioners, about their medical treatment experiences. I have



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talked with many during our annual conferences and through regular email and telephone contact. I believe I am very well placed to comment on the global patient response to Myozyme. To date I know of a small number who are disappointed only to have experienced a stabilisation of their condition (highly valuable though that is), many report great improvements to their strength, stamina and especially in regard to their breathing; fatigue is significantly reduced allowing many sufferers to return to work or engage in activities long abandoned. In the UK we have a number of patients who have been receiving Myozyme for several years through the early drug trials and since EU approval. Over the years I have met or talked with many of them; below is a small sample of their experiences.

Three infants were included on the early trials sponsored by Genzyme, the youngest of whom is now nearly six years old. These children would have died within their first two years of life, however with Myozyme the youngest has just started school and she is already taking swimming and ballet lessons. She is a very bright girl and I met her recently at our annual conference; although not for long as she sped quickly away from me towards the playroom! The two older infants were not diagnosed early enough to experience such a dramatic improvement, their large muscles were too damaged for a full recovery, but their enlarged hearts returned very quickly to normal size and they now are bright, happy and alert children who should lead long and fulfilling lives. The older boy is due to start junior school this year and has been weaned off 24 hour ventilation through a tracheostomy and now needs only nocturnal non-invasive ventilation.

My son was enrolled on the mini-LOTS trial for juveniles at the age of fourteen, one year after an extensive operation to correct the severe scoliosis in his spine. He has been receiving Myozyme for over five years now and is in very good health. His lung function had been deteriorating rapidly and had been using nocturnal ventilation for several years before treatment. His lung function responded immediately to Myozyme and he is now able to sleep safely without breathing assistance. Now 20 years old, Jamie continues with his tennis playing and he is well able to compete with his peers in a number of sports and physical activities.

An elderly gentleman in England has recently reported astonishing improvements in his health after only six months of treatment. He has a tracheotomy and is a wheelchair user. His wellbeing has been restored to the extent that he and his wife were unable to come to our conference this year – they were taking their first holiday for many years! John Shaw is featured on our website (www.pompe.org.uk) under the “News” section

Michelle, a 35 year-old mother of two, was so poorly that she was spending 6 months of every year in hospital with life-threatening respiratory problems. She was enrolled onto the Expanded



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Access Program about five or six years ago. After treatment she was able to spend valuable time without her ventilator and was able to perform a small jump in the air. With time she progressed to using her wheelchair mainly as a carriage for her ventilator. She regained much of her former dignity and was pleased that she could once again stand in her kitchen to prepare meals for her children.

Myozyme was approved in England over three years ago and all of the physicians in our LSD expert centres have reported to me that they are very pleased with the responses they are seeing. We currently have over 90 patients receiving Myozyme in England. Many physicians, who had previously been sceptical about the predicted response in adults, are now enthusiastic supporters of the therapy.

If the Australian regulators require further proof of the beneficial effects of Myozyme they should look out for the case studies that are being reported by treating physicians all around the world.

There are many wonderful reports of the benefits of Myozyme, but the Australian regulators should remember that the most important feature of the treatment is that the patient's condition is stabilised; the progression of disease is halted and life is dramatically extended. One cannot imagine the fantastic benefit that has on the quality of a patient's life. For that reason alone I hope that the Australian regulators will consider your request for approval, so that all may enjoy the full benefits of Myozyme in the knowledge that his life-giving treatment is secured for the future and fully supported by the Australian Pharmaceutical Benefits system.

With Kindest Regards

Allan Muir

Chairman, International Pompe Association www.worldpompe.org

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