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Australian Pompe's Association

5th November 2014

Australian Pompe's Association

Dear committee,

Re: Review of the Life Saving Drugs Programme - submission from the Australian Pompe's Association (APA)

Thank you for addressing the important issue of the Life Saving Drugs Program and its functioning. The Australian Pompe's Association (APA) is grateful for the opportunity to provide input to the Department of Health's *Post-Market Review of the Life Saving Drugs Programme (LSDP),* which we believe will directly impact the future health of our members.

The Australian Pompe's Association represents the interests of the 33 Australians known to be living with extremely rare, debilitating and life threatening Pompe disease. This includes three infantile onset, the oldest of whom is now 10 years old, and 30 juvenile or adult onset patients. Our members include a number of children who currently do not have access to treatment as they were diagnosed over the age of 24 months.

The Life Saving Drugs Program (LSDP) is an essential support to Pompe patients in Australia. It currently provides life-saving treatment to those diagnosed before the age of 24 months with infantile onset Pompe disease.

It has been a great cause for concern to our members and organisation that all people in need gain access to treatment. The PBAC process for the LSDP has been a challenging one for patients to navigate. The APA believes that the review of the LSDP provides an opportunity to address this inequity for Australians impacted by this ultra-rare orphan disease in desperate need of assistance.

About Pompe disease

Pompe disease is an ultra-rare autosomal recessive disorder of glycogen metabolism caused by deficient activity of lysosomal acid maltase Alpha-glucosidase. This enzyme deficiency leads to muscle breakdown, causing disability, which further leads to respiratory failure and in infants, cardiac arrest. Pompe disease is a progressive spectrum disorder, with onset dependent on the amount of enzyme naturally produced. Those diagnosed earlier in life experience faster progression of the disease and a far worse outcome.

There is currently one TGA approved treatment, and enzyme replacement therapy (ERT), Alglucosidase alfa (Myozyme®). A number of patients access treatment through an International Compassionate Access Program (ICAP) run by the manufacturer, however this program was closed to new patients two years ago. One other ERT is also currently in Phase II/III clinical trial BMN701 run by BioMarin for a new investigational compound.

We believe that Pompe disease is the exact type of ultra-rare condition that the LSDP was established to support patients of. Researchers note: "Because of its low frequency of approximately 1 in 40,000 births and the broad ethnic spreading, Pompe disease is a true orphan disease..."[i]

Administration of the Life Saving Drugs Program

The APA is very grateful for the treatment of infants diagnosed with Pompe disease through the LSDP, however we have been concerned about the process for gaining access to essential treatment for those diagnosed after the age of 24 months.

Seven submissions have been made by Genzyme, Myozyme's manufacture, for further access via the LSDP. We understand that the collection of data and evidence in the case of ultra-rare diseases is challenging. It is critical that the LSDP remain a program to provide treatment and funding for ultra-rare diseases that struggle due to their diverse nature and limited data to provide the evidence requested by the PBAC.

39 year old mother, business owner, and community member diagnosed with Pompe disease at 37

"The patients? We are simply stuck in the middle. We are the losers. We have had to become researchers and meet with politicians.

It has become a second job to many of the APA committee members to beg the government to save our lives and it is just unfair. From the view of a patient support group, our focus should be on supporting our members with the struggles that they are faced with in living with Pompe disease.

From a personal perspective, I should be spending time with and focusing on my family, putting all of my efforts into running a very successful family business and focusing on my health and wellbeing."

It has taken a significant toll on our members trying to understand and represent themselves within the system, and we welcome the fact that this is now being reflected on by your committee. While we have been told that the LSDP is not assessed on price on the one hand, we have regularly been told that this decision is indeed impacted by price – something we have no control over. Some of our adult onset members have been receiving treatment now for over seven years and all we really know is that treatment is working for and needed by our members.

At the last review of the LSDP in 2009/10 a number of changes were made. In particular, we are concerned by reports that changes to Criterion 4 have made it extremely difficult for treatments to meet the subjective criteria, particularly when dealing with spectrum disorders. We hope that this review can return the LSDP to its original intent in providing treatment to those impacted by serious ultra-rare diseases.

One recommendation we have is to update the wording of Criterion 4 to remove the subjective word 'substantially.' It may provide more clarity to put a quantifiable goal for life extension, such as an expectation that life may be extended for two or more years.

One of the great challenges we have experienced in appealing for access to treatment within the current LSDP framework is the apparent distinction made between 'life saving' and 'life extension' and how much weight is given to quality of life. We very much welcome the focus on this in discussion of outcomes in your Review of the Life Saving Drugs Program.

Quality of life and survival are intrinsically linked. The need for a wheelchair and ventilator, and also a low Rotterdam Handicap Scale (RHS) score are associated with higher mortality in people with Pompe disease[ii].

In a recently survey commissioned by APA of a nationally representative sample of over 1,000 Australians of voting age we were very pleased to see that they too recognised the importance of quality of life in assessing treatments for funding, and support government funding of rare disease treatment even at high costs[iii]: More than 90% report that they

believe quality of life – such as staying out of a wheelchair or improving breathing – should be a key consideration as well as the amount of time it will extend someone's life. Significantly, more than 85% would support the government funding for medications costing $100,000 to $400,000 per year for a disease impacting 10 to 100 Australians.

22 year old university student was diagnosed at the age of 17 and has been receiving treatment through ICAP for 2 years

"The treatment has enabled me to continue with nearly a normal life... I really need the government to subsidise this treatment so that I am guaranteed a better quality of life and a full life expectancy. Each day I live with the fear that my treatment may halt and therefore my family and I will be faced with the fact that I may not be able to walk and breathe without support. So I plead to the government to give every consideration to helping other young people be given the best chance of a normal life and to get treatment early before the disease progresses.

Without treatment my symptoms will increase and impact the life of myself and my family. I will miss opportunities to marry, have children and maintain employment. My parents want me to have a normal life and with the treatment it enables this, without it, it is not the case."

International comparison

Treatment for Pompe disease is currently significantly more accessible under the healthcare systems in other countries. Treatment was first funded in 2006 in the Netherlands and then shortly thereafter in the United Kingdom and is now available in more than 50 countries around the world, and this has been a great challenge for Australian patients to understand.

This means that there are strong guidelines and histories of programs for treatment in other countries from which Australia has the benefit of drawing, such as those in other well established programs and countries as in Canada and Switzerland.

One recommendation we have that could provide further benchmarking for the program would be for the Department of Health to undertake an annual review of treatments for rare diseases available under a comparable system – the UK's National Institute for Health and Care Excellence (NICE). This could be developed into a comparative report for guidance in Australia.

Clinical effectiveness, the benefit of treatment outcomes and quality of life

44 year old mother and business owner, was diagnosed at 41 and has been on treatment via BioMarin's clinical trial for two years

"The last 6 years have been a roller coaster ride ... To have had the opportunity to travel overseas and meet people who have been receiving ERT and hear their stories of how ERT has stabilised their breathing and walking, coming from an incorrect diagnosis which was untreatable to a correct diagnosis that was treatable felt like winning the lottery!

Then came the news that as an adult we could not gain access under the PBS... and they say we're 'the lucky country' "

We are very pleased that many of our patients can share direct insight and first hand examples of its benefits of treatment for their quality of life and in keeping them alive with their families and communities.

37 year old mother of two young girls was diagnosed at 27 and has had access to Myozyme through the ICAP for 5 years

"Treatment means everything to me and my family. It allows me to be as active as I can as a parent with my children. It gives me the freedom to breath independently and function as normal as possible. It gives me the strength to be as physical as my body allows me to be, it allows me to be here to be part of my family. It allows me to not be completely restricted to fulltime carers and confined to my home. My treatment is giving me life at a stable, healthy, manageable rate that will allow me to hopefully have a full life with my family watching my children grow and develop. Without my treatment life would be a time bomb for me and my family…

Since starting my treatment I no longer need any respiratory support at all. My breathing is significantly better, I function 100% better in my daily routine. My risk of serious injury has reduced, my treatment allows me to walk unaided and for longer periods. My heart has less stress due to my body functioning at a stable level. I am able to function as normal as I can, which is important for me."

Our members have direct experience of the benefits of treatment here in Australia:

- Three infantile onset patients receive access to Myozyme through the LSDP. Without treatment these children would not have survived
- 19 people aged 21-70 currently rely on the International Compassionate Access Program (ICAP) which is run by the manufacture for access to Myozyme. This program was closed to new patients 24 months ago.
- Two people are currently on Phase II/III clinical trial BMN701 run by BioMarin for a new investigational compound.
- Nine patients currently have no access to treatment, including three children.

As stated above in discussion of the administration of the LSDP, quality of life and survival are intrinsically linked.

Treatment provides a wide range of direct and indirect benefits to patients, their families, and the community, including: improved respiratory function, preventing decline and respiratory failure, improved mobility and independence, prevention of cardiac arrest, decreased disability and increased workplace participation and continued interaction with children and the community.

In addition to ERT, many Pompe patients, particularly those whose condition progresses before they gain access to treatment, require a range of devices and supports including breathing apparatus (such as VPAP BiPAP and CPAP machines), walking aides and wheelchairs, modified cars and accommodation.

Patients are aware that their condition, in conjunction with ERT, can be helped by exercise programmes and special diets and supported by information supplied by the Pompe community are working with their dieticians and physiotherapist to ensure that they are doing as much as they can to fight the disease.

a 30 year old single father to his 6 year old daughter, was diagnosed at age 12 and has been accessing treatment through the ICAP for five years

"Prior to receiving this life saving treatment I was dependent on a walking aide and breathing machine at night. I was barely able to maintain a job and was looking at going into a wheelchair permanently.

This drug means my family doesn't have to see me slowly die and waste away from the happy outgoing person I am, and the medical and emotional burden on my family plus the government. Without Myozyme I would not be the fully productive adult I am, that pays taxes, votes and looks after my daughter full time. It means my daughter won't lose her father.

What difference has treatment made for me and my quality of life? It's given me my ability to live, breathe and not be in pain or fear wondering is this my last day?"

first time grandfather at 63, was diagnosed at 58 and has had access to treatment through the ICAP for five years

"I retired several years ago because of poor health which was later found to be Pompe disease. I was first diagnosed after my health deteriorated to such a low level that I was placed in the Intensive Care Unit at Wesley Hospital Brisbane.

After treatment commenced my health returned to a level so that I was able to return to work, begin going to the gym and lead a normal life.

I have regular breathing and blood tests and check-ups with my GP with no deterioration in my condition being noted."

Data collection

APA members are very happy to support and provide data to any studies requested. To date, our members have been submitting data to both the Pompe Registry and the Erasmus Survey international observational studies for up to 10 years.

We recommend standardised testing through a national rare diseases registry. One way of facilitating this would be through establishing a Centre of Excellence in each state for assessments.

Conclusion

The original intent of the Life Saving Drugs Program was to provide access to treatments for ultra-rare conditions to Australians. We hope that this review will return the system to this purpose. One suggestion is to event update the name to the "Rare Diseases Program" acknowledging the challenges – including in data collection - that are unique to rare diseases.

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[i] Gungor, D et al, 'Survival and associated factors in 268 adults with Pompe disease prior to treatment with enzyme replacement therapy', Orphanet Journal of Rare Diseases 2011, 6:34

[ii] Gungor, D et al, 'Survival and associated factors in 268 adults with Pompe disease prior to treatment with enzyme replacement therapy', Orphanet Journal of Rare Diseases 2011, 6:34

[iii] Australian Pompe Association data on file – a survey of 1,003 Australians over the age of 18 was undertaken in July 2014 by independent research agency PureProfile commissioned by the APA