# SUBMISSION BY THE AUSTRALIAN POMPE'S ASSOCIATION

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To

# SENATE FINANCE AND PUBLIC ADMINISTRATION REFERENCES COMMITTEE

# INQUIRY INTO THE GOVERNMENT'S ADMINISTRATION OF THE PHARMACEUTICAL BENEFITS SCHEME

Thank you for the opportunity to present our submission to the committee.

(b) any consequences for patients of such deferrals;

The Australian Pompe's Association is most concerned that the Cabinet should have the final decision on treatment. The members of the Cabinet have no knowledge or experience of such rare diseases as Pompe disease with only 0.00010% of the Australian population.

Most doctors have never seen a Pompe patient before, how can a dedicated group of busy politicians understand the challenges and needs of 22 patients who are located throughout Australia.

The Australian Pompe's Associate believes it is too easy for the government to reject something it does not understand, affects so few and has no experience of.

The Australian Pompe's Association (APA) represents 22 patients in Australia who suffer from adult onset Pompe's Disease. Pompe's Disease is a very rare, progressive, fatal, genetic, lysosomal glycogen storage disease, which happens when both parents pass down to their child a copy of the faulty gene which is responsible for Pompe's.

Pompe's Disease is caused by a complete or partial deficiency of the enzyme acid alpha glucosidase. This enzyme is responsible for breaking down excess glycogen which accumulates in the lysosomes of the muscles. As the Pompe's patient is deficient in this enzyme, the excess glycogen is not broken down and continues to accumulate in the muscle cells. This results in progressive muscle damage and severe muscle weakness, so that normal muscle function is increasingly impaired.

Pompe's patients struggle initially struggle with normal daily tasks, such as walking, climbing stairs, getting up from a chair, eating, swallowing, breathing, combing hair, cleaning teeth, showering, using the bathroom or just turning over in bed. Things become more and more difficult to do as the disease progresses and patients finally succumb to their illness. Since July 2008 three of our APA members have passed away due to their Pompe disease, which is 15% of our membership.

Pompe Disease can present itself at any age from birth to older adults, its severity often depending on the age of onset, and level of enzyme activity.

Babies have the most severe infantile form of Pompe's Disease, and can develop symptoms in the first few months of life. These symptoms include muscle weakness, feeding and digestive problems, enlarged liver and heart, enlarged tongue and considerable respiratory problems, due to the total lack, or a severe deficiency of the enzyme 'acid alpha glucosidase'. The infantile form of Pompe's Disease may progress very quickly and babies born with this form without treatment usually do not live long enough to celebrate their first birthday.

Over the last ten years Genzyme Corporation has developed a treatment for Pompe Disease. This treatment is an Enzyme Replacement Therapy, where the patient is infused, every two weeks, with Myozyme (alglucosidase alfa), a manufactured enzyme.

Myozyme has proved to be a great success overseas and over 44 countries worldwide have funding available for Myozyme. To provide treatment for the seriously ill Pompe patients in Australian the manufacturer Genzyme established a temporary compassionate access scheme to allow Australian Pompe patients treatment while TGA and PBS approval was undertaken.

The first patient stated treatment with Myozyme under the compassionate access scheme in 2006 and then a further 6 in 2007. In March 2008, Myozyme received marketing approval from the Australian Therapeutic Goods Administration, for the long-term treatment of patients with a confirmed diagnosis of Pompe Disease (acid alfa-glucosidase deficiency). In 2010 the compassionate access scheme was closed to new patients due to the ongoing delays with PBAC approval.

Currently there are 22 known cases of Adult onset Pompe Disease in Australia with only 16 adults receiving treatment under the compassionate access scheme. The distribution across the country is as follows.

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|---|---|
| New South Wales | 9 |
| Queensland | 6 |
| Victoria | 5 |
| Tasmania | 3 |
| South Australia | 0 |
| Western Australia | 1 |

Since July 2008 five submissions have been made to the PBAC the last being in July 2011. Each time a submission is rejected the patients receiving treatment are faced with the possibility that their treatment under the manufactures compassionate access scheme will be withdrawn and their disease will return to its normal progression. The patients who are currently awaiting treatment are once again disheartened as their disease continues to progress. This constant rejection is very troubling for our members as they a faced with the major decision of life to have a family or to buy a house knowing that their treatment made be withdrawn any day or never be started.

Myozyme is a very effective treatment but it is only able to stop the progression of the disease it is not able to correct the years of damage that the patient's disease has caused. The most important thing is to treat early before more damage is done.

Should additional delays be caused to the drug approval process our members who are currently not receiving treatment will progress more and faster. Continued delays may well result in the closure of the compassionate access scheme causing the 16 patients to once again be subject to the unrelenting progression of their disease.

Myozyme is the first in a generation of Enzyme replacement drugs but for our members to benefit from the second and third generation drugs they need to be alive and be treated now without further delays.

The APA believes that it is critical to treat Adult Pompe's patients as soon as recommended by their specialist, while they are still in the initial phase of the disease. This will ensure that patients are able to maintain employment and remain as productive members of the community. Many of our members would have been diagnosed much earlier if the current diagnostic capabilities had been available when they first experienced symptoms.

Such rare diseases as Pompe disease with 0.00010% of the Australian population is hard for an expert body as the PBAC to understand. Requiring the health minister and Cabinet to assess the need for urgent treatment is challenging. The APA believes it is too easy for the government to reject something it does not understand, affects so few and has no experience of.

Helen Walker OAM
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20th July 2011