

## INFORMAL SURVEY of Australian POMPE PATIENTS – since starting Treatment with MYOZYME



This is an informal survey of Australian patients who suffer from Pompe's Disease, to show the effects of treatment with Enzyme Replacement Therapy – MYOZYME.

### The Australian Pompe's Association

July 2009.

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#### POMPE'S DISEASE –

Pompe's Disease is a very rare, progressive, fatal, genetic, lysosomal disease. The effects of Pompe's Disease are progressive muscle damage and severe muscle weakness, so that normal muscle function is increasingly impaired. Pompe's patients struggle with the most normal daily tasks such as walking, swallowing, breathing, using the bathroom. Just cleaning teeth and combing one's hair can often be a difficult task. All these ordinary things become more and more difficult to do as the disease progresses. Most patients will have to use a wheelchair, which will limit them even more.

Respiratory muscles are also very much involved, affecting pulmonary function and, without treatment, most patients will have to use a ventilator. A simple cold can have dire consequences for the Pompe's patient as it can quickly result in chest infections, pneumonia and death.

As the disease progresses the patient can become bed-ridden, have to rely on tube-feeding, have the need for a tracheotomy with ventilation, and need 24-hour care.

Often the caring is done by a loving partner or parent, son or daughter.

Pompe's Disease is a 'shared' disease, as it does not affect only the patient but, in fact, affects the whole family!

Pompe's 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.

The 'infantile' form of Pompe's Disease will progress very quickly and these babies may not live long enough to celebrate their first birthday. They will require urgent life-saving treatment with Myozyme.

From 2004 until now, at different stages, Australian patients have started treatment for this devastating illness. Following, we show the results of an informal survey where these patients report on the effects they have noticed since they have been treated with Enzyme Replacement Therapy – MYOZYME.

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#### Australian Pompe Patients

Of our patients: 4 patients have died (aged from 4 months old to 49 years old)  
1 patient does not want to have treatment

#### At this time we have 19 adult Pompe's patients who do need treatment

Of those – 14 patients were surveyed, and from those 14 we received a 100% response rate.  
2 patients have only just started treatment and did not participate in this survey  
2 others are still waiting for treatment and did not participate in this survey  
1 patient is presently overseas and did not participate in this survey

#### Surveyed Patients' Age and Treatment Range:

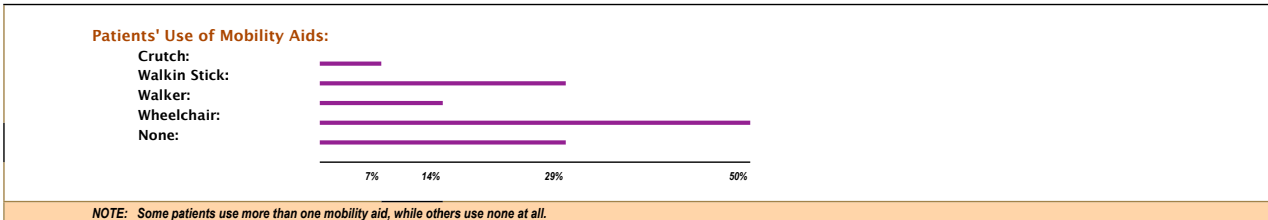
age at diagnosis      age at start of treatment      period of treatment to 1st July 2009

12	24	1 y 1m
16	29	2y 2m
26	39	1y 5m
26	49	1y 4m
36	67	3y 5m
37	60	1y 11m
40	63	2y 3m
41	44	2y 1m
42	53	3y 3m
42	48	2y 1m
45	50	1y 4m
50	67	1y 11m
51	56	2y 4m
57	58	3m

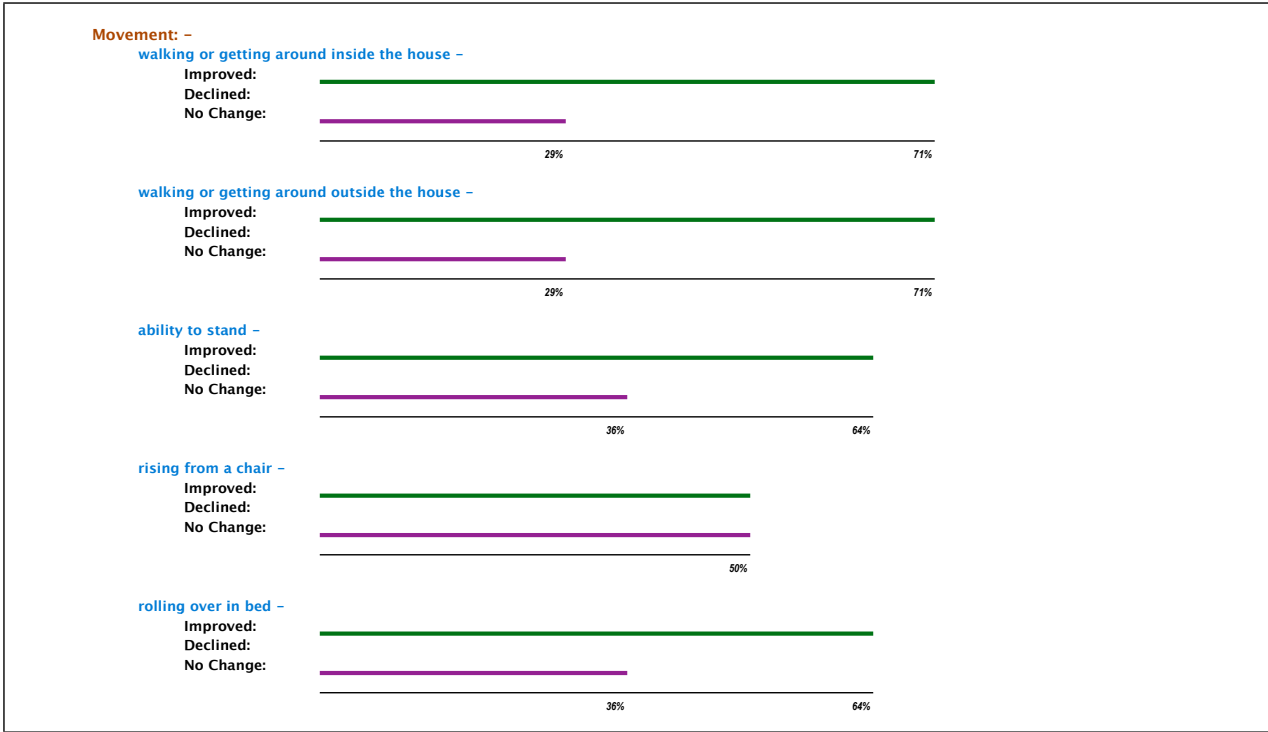
As shown in the above table, the ages at the start of Myozyme treatment range from 24 to 67 years old, with the average age being 50 years 6 months

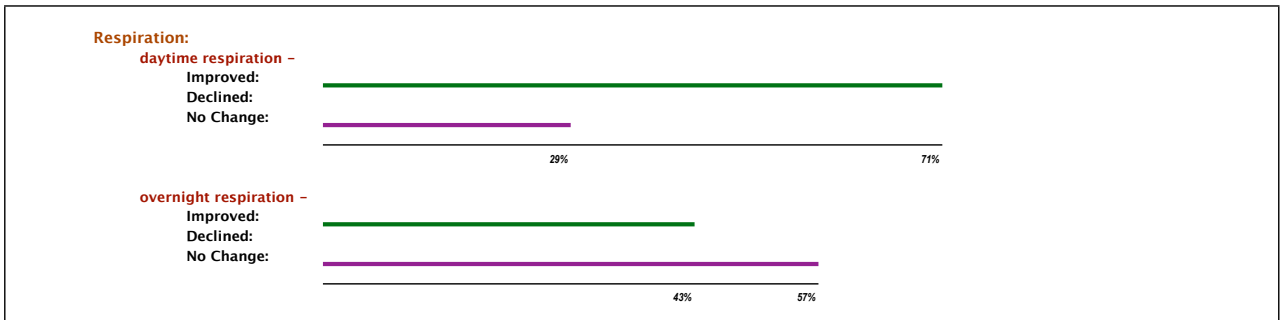
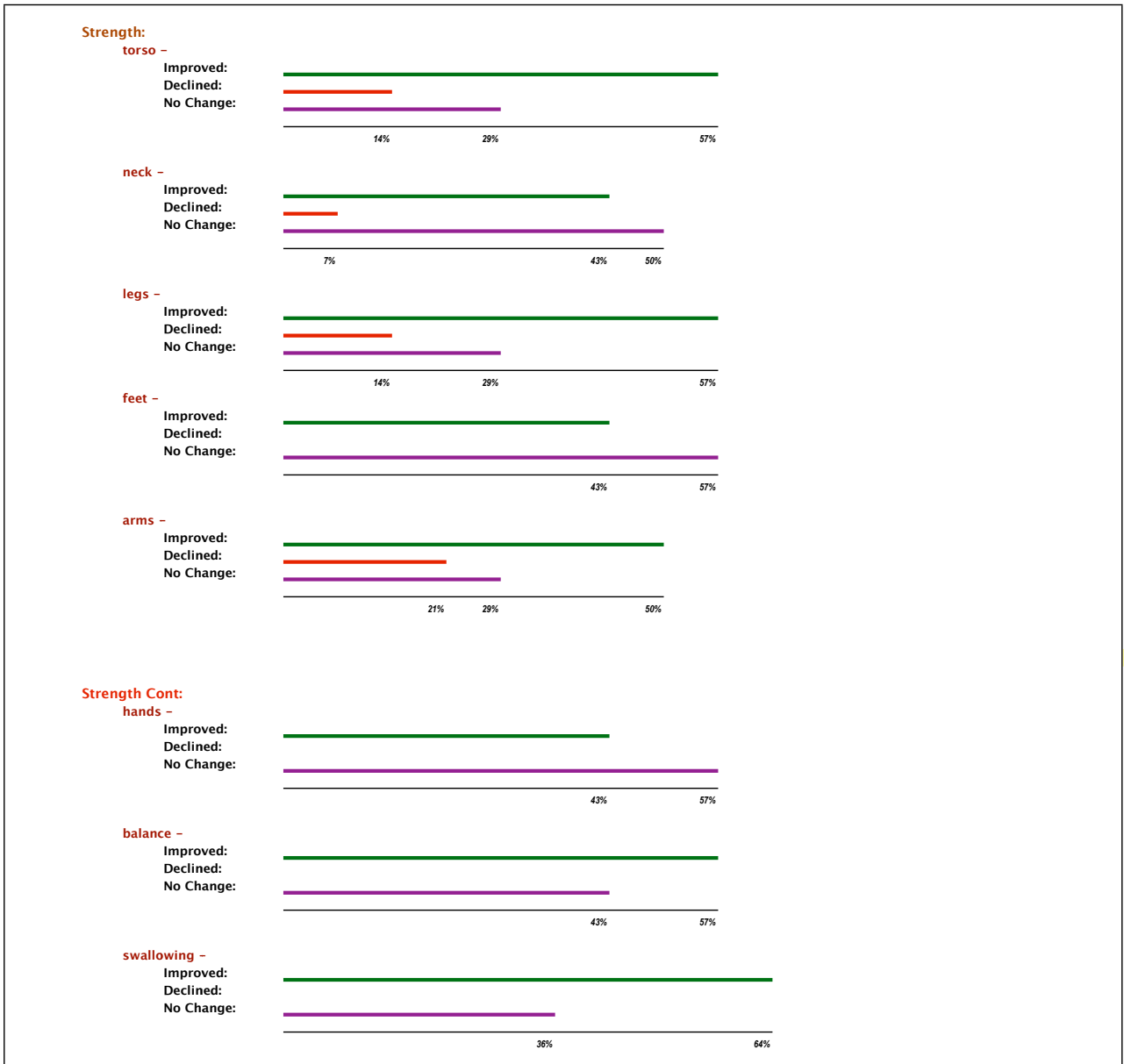
The period of treatment ranges from 3 mths to 3 yrs 5 mths, with the average period being 1 yr 11 mths.

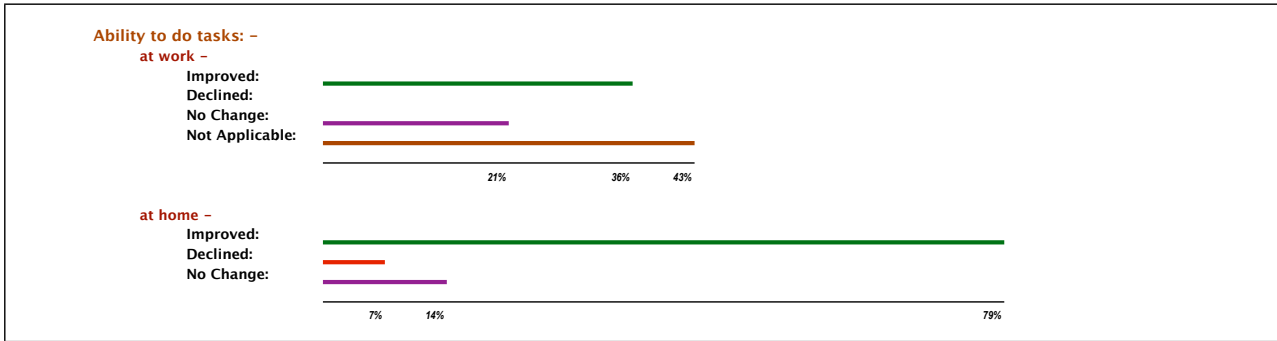
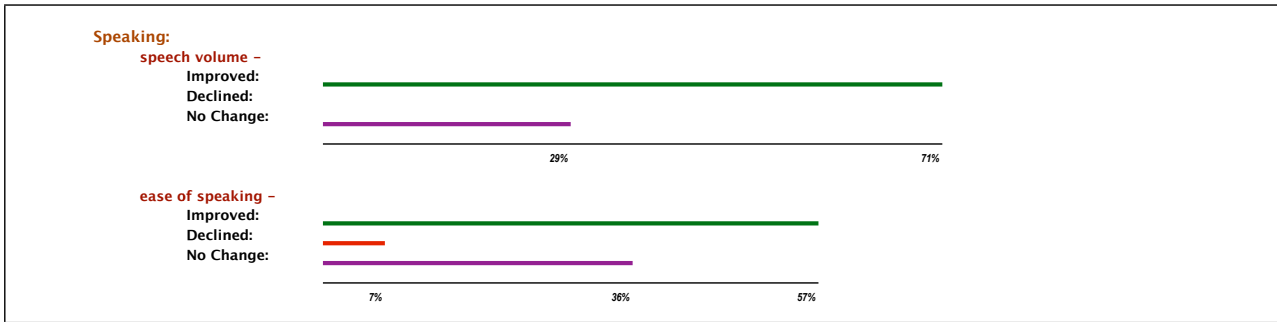
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**NOTE:** Before treatment Pompe patients continually deteriorate, some at a fast rate, others at a slower rate.  
 In the sections that follow you will find a category 'no change'. While 'no change' means 'no improvement', it also means 'no further deterioration' or 'stabilized'.  
 As this survey shows collected data since patients started treatment, we should consider 'no change' (or 'stabilized') to be a plus in itself.







**SUMMING IT UP –**

As was mentioned before, 'no change' will mean that there is no improvement but, also, it means that patients have stabilized, and have not lost further ground. As Pompe's Disease is a progressive illness, 'no change' or 'stabilized' should be considered a plus.

As shown in **Surveyed Patients' Age and Treatment Range** on page 3, many patients had a long wait from diagnosis to treatment, with the associated deterioration and resulting damage to their bodies. Yet some of them managed to work at a job, and most raised families, albeit with difficulty and some very trying physical circumstances.

It should be remembered that most of us patients measure our strength by how well we can walk, work, push our wheelchairs, or just breathe on any given day, and how well we can live our everyday lives, doing everyday things, working, raising our families, and speaking without becoming breathless or needing a ventilator to help us breathe.

And – very importantly – living a life that is independent of help from others.

All the 'normal' things that most people just take for granted.

When we can perform better in the workplace, better raise and be involved with our families, and when we can live our lives better by doing the ordinary everyday things better, then we can truly say "Yes, that's improvement!"

Had treatment been available to us at the time of diagnosis (see table on page 3), we would not have suffered the devastation that is Pompe's Disease, with its always progressing weakening of muscles, loss of functioning and compromised breathing – and we would not now need the care of our partner, parents, siblings or children. Or worse, be placed in a Nursing Home at a much too early age.

Not only does that bring much emotional trauma and heartache to the patients and their families, but it will also bring with it considerable costs to the Australian Government for many years to come.

Pompe's Disease is a devastating, progressive, fatal illness that can be likened to a cancer that goes through the body, to destroy our muscles, our strength, our functioning, our breathing, our soul.

It is unrelenting and ongoing and will quietly, but surely, continue to do its damage until death, or until something is put in place to stop this progression.

For us, the Pompe Patients, that something is Myozyme!

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**SUMMING IT UP – Cont.**

As early diagnosis is now possible, Myozyme treatment should be started as soon as diagnosis is made, and well before Pompe's Disease has a chance to run its destructive path, so that Pompe patients, also, can live their lives to the full, independent of help from others, or nursing care facilities.

Life is precious, and living a life being sick, and dependent on others, is a huge price to pay.

We hope that this survey will be of help to those who read it, and will give some clarity as it is patient orientated. In closing, we would like to say a very sincere "Thank You" to all who took part in this survey.

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**The Australian Pompe's Association**  
June 2009