

## LIVING WITH POMPE'S.

by Betty Cook.

In 1962, when I was 23 years-old and seven months pregnant with my second baby, I found, one morning, that I could not straighten up from a bent-over position. That lasted a few weeks and the problem corrected itself.

The doctor had no answers.

Little did I know then that thirteen years of misdiagnosis were to follow, and as a result of these misdiagnosis, the wrong medications were given. Medications, I found out later, which were contra-indicated for Pompe's Disease.

In 1963 it became difficult to climb stairs with a toddler on my hip - maybe I needed more exercise.

By 1964 others noticed that I had developed a definite gait, and I looked as if it was painful for me to walk. I had not noticed anything wrong with my walking.

In hindsight, that was to be the first example of how this disease creeps up on one without really noticing it, as it all happens so subtly, and the body compensates without the patient realising that it is doing so.

By the time you do realise, much muscle damage has already been done.

In 1965, again I could not straighten up from a bent-over position. I had to 'climb' up my legs and use props to straighten up. This time the problem was here to stay, and it was to cause great difficulty and frustration with two little ones to look after. Just to bend one's back and straighten up again - we do that movement a hundred times a day without ever thinking about it ..... until we can no longer do it! Then we really find out just how the loss of that ordinary movement limits the activities of daily living. Especially while caring for small children.

I now had to plan and strategise to cope with an ordinary day.

I felt weak in the lower-back and thighs.

I could no longer pick up my children off the floor.

End 1965 we moved to a new house and, with that, a new doctor. My first consultation with him was interesting. I explained the problem I was having straightening up from being bent-over, and also that my thighs and back felt weak.

The good doctor thought that it was all due to my "nerves", told me I was "depressed" and diagnosed me as being "totally inadequate!!"

He told me: "Go home! It's all in your head!"

Some months later I had to see a gynaecologist.

While I was there I told him of my problems.

The gynaecologist could see that there was in fact something wrong and sent me to a neurologist, who sent me to a second neurologist.

To make it short, in 1975, after being hospitalised for six weeks, twice, many tests, EMG's, blood tests, biopsies, etc., and being misdiagnosed, twice, as having Polymyositis, and being treated with Prednisone for 5 years (which is contra-indicated for Pompe's Disease), I was finally told that I had Acid Maltase Deficiency - aka Pompe's Disease.

After thirteen years we finally had the right diagnosis.

Only to be told that there was no cure!

No cure?? What do you mean "No cure"??

That just didn't seem right!

When you are sick you go to a doctor, and you think that he will give you medication or some other good advice, and that will fix you up so that you can go home and get on with your life.

In fact, we expect that!

But there was no cure and no available treatment ..... Not for Pompe's Disease.

The fact that there was no cure or available treatment brought a whole new range of emotions which needed to be dealt with.

The fear! Just how bad was this going to get?

The anger of not being able to fully live life without struggling.

The guilt and frustration I felt, for the things my husband and my kids were missing out on, because of my disabilities.

The uncertainty of what the future would hold for us, and what was yet to come.

The awful loneliness of living with a disease that's virtually unknown to the rest of the population, except maybe your doctor.

The frustration and helplessness, as Pompe's Disease slowly but surely takes control of my physical life.

Having to forget about the plans I had of going back to work when the children had started school. And later, the disappointment of having to give up the Do-It-Yourself projects around the house, which I loved doing so much

By then thirteen years had passed and my symptoms had worsened considerably.

All of this affected my children the most. There were many things that I could not do with them, many activities in which I could not take part. Even just going to a school's Sports Day with them was hazardous. As I got weaker, my balance was badly affected and one small bump from one small child, or some uneven ground, a small pebble, or a gust of wind was enough to make me lose my balance and end with me sitting on my butt on the ground, being unable to get back up.

And often these falls ended up with twisted knees or twisted ankles or sprained feet, and quite a few bad headaches when my head would hit the ground.

Swimming Carnivals especially were very scary - heaps of kids, wet slippery ground .....

I was not alone with this illness ..... my husband, my children and I, we all suffered from Pompe's Disease.

Our whole family was well and truly affected.

But, I don't want this to sound like all doom and gloom. We also had a lot of laughs and a lot of fun and a lot of good times together, despite the limiting circumstances.

Over the years I was to experience many limitations.

In fact, I feel that we - the Pompe's patients - spend our lives constantly grieving over the normal things in life, which we used to be able to do with ease once upon a time, and are now just being chipped away little by little every single day, while we are left looking on helplessly and totally frustrated by the process which is destroying our muscles and a normal life.

Some of the limitations and difficulties doing just simple and very ordinary things:

- Straightening up from a bent-over position

- Getting up from a chair
- Struggling to get up from a toilet
- Struggling to get in and out of a car
- Stepping from a road onto a footpath
- Climbing stairs
- Making beds
- Doing the normal household duties that everyone just takes for granted
- Rolling over in bed
- Hanging out clothes
- Excessive tiredness
- Affected diaphragm and breathing muscles, so that serious breathing problems result.

Just balance was a big issue.

I lost my steady balance when standing and walking and had to be very, very careful and constantly aware of every step I took. Every step could - and often did - result in a fall, with painful consequences. This would mean that I was unable to walk at all for weeks at a time, and would have to use a wheelchair. It would also mean regular appointments with the physiotherapist, with associated costs.

Every fall took away a little more of my confidence and made me just that little more wary to get up and try again.

There have been many times when I have had a fall during the day, and I would sit on the floor and wait for someone to come home so they could help me up, as I was unable to get up off the floor by myself.

Until the day came that the fight to stay on my feet just became too much and I was to be permanently in a wheelchair. There had just been too many falls, too much pain, and how many more times did I really want to bang my head against the floor?

A few years after that I also had to start using a ventilator.

This disease isolates patients simply because it is so rare. That makes this illness an unbelievably lonely experience.

No one to just talk to, who would understand just what you are talking about and what you are feeling and what you are going through.

You have to deal with doctors who cannot really answer your questions, because they don't know the answers and, often, will not take your complaints seriously.

Being a 'one-of' patient does not help matters.

Finding that health professionals in most categories are at a loss, because in their particular textbooks there is no mention of "Pompe's Disease".

Or on the other side, the health professional who pretends to know it all, while you know yourself that that isn't so.

You know it, because you are living it.

It was to be 33 years, in 1995, before I got to know someone else with Pompe's Disease.

Over the years when my children were at school I missed out on many school activities like sports days and carnivals, parent/teacher days, because there were just too many kids around. Recreational activities with the kids also were limited to the kids doing the activities and me looking on.

I feel that my kids missed out on a lot of things, because of this lousy disease.

Just the ordinary everyday things, that are so taken for granted, but yet so very important in the child/parent relationship.

Now my boys have children of their own - my grandchildren. They are growing up with me being in a wheelchair and are very accepting of that. When I am invited to attend something from their school, these little kids now vet out the terrain first, to make sure that the wheelchair does not come up against obstacles. Very matter-of-fact I am told which entrance to use " ..... 'cause there's no steps there!"

I am very, very lucky that I have a great husband and kids, family and some good friends. My husband especially, and my boys deserve a medal for their understanding, and the care and help and encouragement they give me so unselfishly.

Now I can no longer stand.

It takes me (with help)  $\frac{1}{2}$  hour just to have a shower and dress in the mornings instead of 10 easy minutes .....

I have to use a lifter to get into a shower-chair, to toilet, or to get into / out of bed or wheelchair.

Just to go shopping or to go out for a while needs planning in advance.

Little seems to happen spontaneously now.

After many years, being forced into a disabled lifestyle, with its many limitations and frustrations, is still a very emotional issue.

Outings with family and friends become less and less as these things just become too daunting to attempt. Often invitations are refused because it is just too difficult and mostly impossible to get around the 'must-do's' whilst attending, even with an assistant to help .....

Just toileting is a major hassle and often pretty well impossible. So most of our socialising with family and friends is done at home.

So I now live in a world which is not really designed for disabled people, which brings its own headaches.

Being in a wheelchair in a country which is decidedly wheelchair unfriendly is a serious business and not at all funny. Much improvement is necessary here.

Pompe's Disease is a horrible, insidious, limiting, debilitating and frustrating illness, which will creep up on you without you realizing the damage it is doing, until much later. It will totally up-end one's physical ability and with that, often, one's dignity, self-esteem and peace of mind.

It will rob you of your self-confidence when you need it most.

## Enzyme Replacement Therapy

Enzyme Replacement Therapy to treat Pompe's Disease has become a reality. I have lived with Pompe's for 43 years now, and it couldn't come soon enough.

I have been getting treatment with Myozyme for the past eleven months. And I am so happy that I am being treated.

During the last 6 months of 2005 especially, I really felt that I was going downhill at a very fast rate, and I was very worried.

Now, after eleven months of treatment, I keep getting comments on how well I look, and I am told that my voice sounds noticeably stronger. Also, last year, if I had to make a phone call that I knew would last more than a few minutes, I would go on the VPAP while I made that call, otherwise I would just not have enough air. I rarely do that now.

My breathing has improved a little, I have more energy, better colour, less slumping of the shoulders, better outlook (*well of course* ☺), stronger voice, a very good appetite, a little more stamina and I feel just great!

And while I cannot really see any difference from one infusion to the next, I can see - looking back over the past year - a big difference to what I was twelve months ago. I feel and am much better. I certainly have not lost any ground.

Life is good and I am very happy to be treated.

Illness, when you have had it for a very long time, changes things so that, in time, it is no longer experienced as an illness by the patient. It becomes so much a part of your life and being, that it is experienced more as "normal" life. Now it's just so good to actually feel better!

Many thanks to all who have worked so hard towards getting treatment to the Pompe's patients.

Thank you so much!

*December 2006.*

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