

DYING ALONE BECAUSE OF POMPE'S DISEASE

HELEN

BACKGROUND Single - one living brother who lives interstate, approximately 2300 kilometres from me. He has heart problems. There are no second-generation children.

I could start my story with the usual noted facts of what happened as a child because of limited physical ability or I could relate to the loneliness of not being able to participate in sport while living in a sport fanatical country of outside activities.

I choose another point of concern that is not often talked about that is the dictatorship of Pompe's disease in your journey towards the end of your life.

While I was fortunate to have a so called normal life as a child I continued into my early twenties until the first symptoms started to show that all was not right. I had my own business but worked with several associates who were there for me as the days became more difficult to work.

My work was in a financial section of the commercial world.

Being self-employed put pressures on me there were no back up systems to pay sickness payments.

However this position allowed me time to be involved in many community projects in a provincial town of 10000 in Australia, I could never take a position of total commitment, because I never felt I was sure of what my health would be on the designated day of involvement.

I always worked in a group knowing that I must have a back up if I was not fit.

While this put restrictions on what I could achieve I did manage to accomplish positive results in a number of projects that helped my fellow citizens.

As I was approaching the peak of my community involvement I was very aware that my health was now deteriorating at an alarming rate.

I had searched for medical advice for so long and had 5 stays in hospital for surgery that never did correct the problems of dislocating shoulders. It came to a situation where I was questioned as to my mental ability to cope with the pressures of life. This made me very cautious of how hard I should try to get diagnoses, however I really knew that I was in strife and persisted.

Eventually I was diagnosed with Pompe's Disease at 49. I then worked towards finding another Pompe patient it was the beginning of the Australian Pompe's Association.

My ambition was to get an understanding, perhaps treatment, and have some quality of life that would allow me to retrieve so many of the daily living pleasures I once had.

That was 16 years ago it was a life that had offered so much to the community but that commitment was taken away by an insidious disease that crept through my body on a slow march until it engulfed me with every conceivable problem it could give out.

It deceives the public by its so called "you look well" assessment. This has much the same results for the professionals that attend to you for some time, but as it unleashes its fury on your body it starts to convince those close to you what a horrible disease it is.

Over my 15 years in community service I was an active member of my Community belonging to a Professional organization, being an elder of my Church, being on a board of management for an aged care home for 70 residents for 13 years. Being a treasurer of a regional art gallery, Chairperson for a heated pool committee for disabled, Chairperson on a board of management for the physically disabled young adults home.

While this type of community involvement may be a pattern for many people in a provincial town it was unthinkable that this would be taken away from me as I was

only in my forties to me it was such a waste of human endeavour because I was to trying to help others by contributing to the community.

New challenges are presented to the Committees but you are no longer able to be there.

Not even can you contribute by picking up the phone and talking through an idea ---
- this is gone because talking becomes an issue as your diaphragm is so weak that you cannot get a breath ---there is no way you can speak with any volume---- your tongue is so swollen that pronunciations of words fail. Saliva often dribbles from the corner of your mouth ---- you dare not try to drink unless the drink is thickened. The thought of uselessness over takes you. The visit of colleagues starts to diminish, as they are involved with the latest challenges of your community. Some friends shift from the district and many no longer can face the distressing sight of someone that is obviously struggling with a disease that they know little of. They now have nothing in common to talk to you about.

This is nothing new you say this is what happens as anyone ages.

But I say not at 40 when most of your mates are still around.

Normal old age barriers appear as friend's age with you and society is programmed to cope with that, but not with this Pompe's disease at an early age.

Because it is so rare explanation to people is left alone as the fear of "oh no not her again with her problems we know nothing about it its too hard to understand ---- we can do nothing about it "

It becomes as boring to them as the general discussions of a group of arthritics complaining about this ache or pain.

And so the only action is retreat as the difficulty of talking because of muscle weakness takes over, more and more you have to use the respirator just to get through the day.

So many other Pompe patients will give you an insight to that problem.

But my heading of dying alone, because of Pompes becomes a reality one cannot make new friends or mix with the general flow of community life. Isolation becomes the normal day-to-day existence.

No longer can you mix socially, go shopping or just enjoy a ride in the car. No one understands this dreadful disease ---yes family eventually do, but that is not so for me I have none.

Even with the best intentions people who you think you know and understand the situation --- they don't.

If they are not family they are not obligated nor should they be responsible for the well being of you because of this rare disease.

Friends must have their lives to live while they are fit enough to do so

Life is short for many --- Life is for living --- Life is not meant to be this terrible lonely struggle that could be helped if the Enzyme Replacement Therapy was approved for use.

So much could be given back to communities so much could be saved.

So much enjoyment for so many.

I do not want to die alone because of Pompe's disease.

Helen

Australia



Helen



Gardening, something I love