

My Case Study

Robert Charles Gooch

Australian but now living in Los Angeles for the LOTS trial



I was first diagnosed in 1983, by Professor John Hopwood's facilities at the Adelaide Children's Hospital. My particular genetic mutations have since been defined through the Rotterdam facilities of Doctor Arnold JJ Reuser, Biochemist, Clinical Genetics, Erasmus University, Rotterdam. When I attended the Late Onset Pompe Observational Study in Seattle WA last year in 2004, a further diagnosis confirmed my Pompe condition as part of the study qualifications.

Being diagnosed was not easy. My friends and employees motivated me in the early 1980's to seek an explanation for my funny walk and lordosis. It took several visits to various specialists, with each visit ending in a discussion along the lines that I had nothing wrong with me, and with me answering that I felt they were saying they couldn't find anything wrong and where should I go next to find someone who might. In this process I suffered all sorts of horrible tests assuming a neurological involvement. I remember one test which involved a long needle was inserted into the back of my leg muscles and then being asked to move them.

Eventually a hypothesis for Pompe's was tested, and a huge lump of "sashimi" was taken from my leg and stomach muscles. The "sashimi" description was the surgeons, and it was one of the most painful procedures I have ever had to endure. In contrast, the small skin sample taken in Seattle was painless.

My condition at age 59 is better than most. I have tried to preserve my condition with daily hour long walks and physiotherapy three times a week. I can still walk but with difficulty, and I find myself falling on average once every ten days. Most are harmless with little more than grazes and bruises but lately I have been less able to save myself. In the past year separate falls have resulted in a broken rib and a lacerated scalp requiring stitches. At most times I am carrying some form of injury from falls, such as bruises, grazes sprained wrists, elbows, knees or a stiff neck or back from a jolting fall. Crossing busy streets has now become too dangerous to do on my own without a stabilising hand from a carer. A stick no longer is enough. I am no longer able to carry weights, as just walking now fully occupies my abilities.

I cannot get up from the floor without assistance or climb stairs without a hand rail. I cannot bend forwards to eat, nor get out of any chair apart from an upright one, and then only with a suitable table in place and with people around to stabilise both the chair and table. This makes life difficult; it means having to stand a lot and sometimes for the entire time if no suitable sitting situations can be found. I also now find it impossible to spend any length of time in a seated position as my breathing difficulties cause me to become mentally dull and if I have just eaten I suffer unbearable discomfort. It is as if pressure is put against my lungs and heart.

Since 1987 I cannot sleep without my respirator. This is a common Pompe problem as our diaphragms fail and with sleep we suffer severe sleep apnoea when we depend upon our diaphragms to breath. Untreated apnoea in these circumstances would be fatal over time. When my face mask leaks in the night, then the next day I suffer the apnoea symptoms typical of a bad alcoholic hangover. If I fall asleep without my respirator, then I

wake with eyes wide and my heart pounding in the same way a normal person would if they had their airways blocked. In the past year my breathing has become noticeably worse and my speech slurred as the muscle dystrophy progresses through my body. I find it difficult to speak loudly and often run out of breath before I finish a sentence. The stress of my failed marriage over the past three years has accelerated my rate of deterioration. Something other Pompe patients have noticed in times of stress. I am now not able to live safely on my own and adequately care for myself. More importantly the risk of a serious fall, where I might find myself in a cramped position where I cannot breathe is a reality and a constant fear. This happened once when I fell with my arm under my rib cage causing sufficient pressure on my lungs to stop me from breathing, and being unable to move to save myself. Fortunately at the time my son was near and able to lift me clear. (Writing about my condition with such honesty has not been easy.)

Potential treatment from my participation in the upcoming Late Onset Treatment Clinical Trials couldn't come sooner, though even with this there are no guarantees. I stand a one third chance of receiving a either a placebo, a half dose and a one third chance of the full dose. At the end of the trial there is hope that the FDA will require follow up data, and this would result in me receiving a full treatment.

Pompe's has had its psychological and financial costs as well.

I suspect it was a major contributor to my failed marriage of twenty three years. Going out into the world and living on my own was without doubt one of the most difficult things I have ever done. It was dangerous as well and one fall when on my own required stitching to the back of my head and proved to me that I needed a permanent carer in my life.

Needless to say I was feeling especially alone and vulnerable at the time.

I feel I cope well with the potential depression that affects other Pompe patients. Adversity can bring out the best in some people and I feel it a blessing that I am one of those who can cope positively

Pompe's has had its financial costs as well.

During the late 1970's and throughout the 1980's I developed a sizeable business, worth probably in excess of AU\$20 million at the time. (US\$15 million.) Certainly my chief opposition at the time sold out for four times that amount back in the late 1980's. I had an Australian wide network of investment advisers and financial planners in all Australia's major towns and cities and in all States and Territories, some twenty five offices in total. All this was lost in the late 1980's when untreated sleep apnoea left me unable to think, plan or logically work out what was happening to me. I lost my will to stay dedicated to my business with my diagnosis in 1983 at the age of 37. The doctors wouldn't tell me anything and my research showed the disease was fatal and that I probably had less than a ten year life expectancy. The literature I found was in medical terms and difficult to understand, and apnoea was in those days killing Pompe people off at a much younger age. This took way all my work incentive and left me in the bewildering "why me ?" state of mind cancer sufferers often feel.

The "sick canary syndrome" set in where the healthy peck to sick to death. My senior management grouped together and tried to steal my business. They failed as most of my field advisors stayed loyal, but it did succeed in breaking up my business. The ensuing uncertainty, along with my opposition sensing an opportunity to steal my trained personnel away with financial incentives led to the exodus. I was a pioneer in the financial industry and the changes I caused to the established ways of the past made me some powerful enemies, especially from the stock broking fraternity and they succeeded in putting pressure at the beurocratic level with my security dealers licence. In the end, I was in no condition to want to defend myself and I let the whole disaster collapse looking for an escape from the living nightmare I was enduring at the time.

So Pompe's in a way was responsible for the collapse of my major business worth many millions.