

## Personal Experiences with Pompe's Disease

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### **Age 0 - 10**

In January 1940 I was born as the fourth child of seven. Three sisters had come before me, and one brother, one sister, and then one more brother came after me.

To my knowledge, I was a normal baby with normal development. I experienced my youth as pleasant and safe. Only later in life did it strike me as really odd that in elementary school I did not like sports days, whereas the others experienced them as a rather pleasant break. I liked sports in themselves. Upon analysis I came to the conclusion that the feeling of displeasure was mainly caused by the fact that I did not like to sprint (and that I could not really sprint). Also things like sack races, crawling quickly through a barrel, etc., were activities in which I achieved nothing at all and therefore did not enjoy.

At the age of 8 I joined a gym club and stayed in it for about one month. Then I went in for football, which I kept up for a couple of years because I liked it, though I could not play well. Very curious.

In general, I was not aware that anything was wrong during this period.

### **Age 10 -20**

A period of transition from elementary school to MULO [higher elementary education], followed by dancing lessons, dating, and military service.

Up to my term of military service I had lived an untroubled and normal life and accepted that I could not do certain things but could do other things quite well.

I did not experience the MULO period as problematic in regard to muscular activities. I used to go on bicycle holidays with friends in which we usually covered 100 - 150 km a day, even more on a few occasions.

Through dancing school I began to court my present wife. We have always liked to dance quite a lot.

In the military it first struck me that when I sprinted in sportswear my final time was about 14 to 15 seconds per 100 meters and that when I ran wearing field clothes and pack my time was approximately the same. During the sprint in sportswear I also had the feeling that I had the tendency to drift or have my knees give out. Whereas with a pack my "position" was more stable. Even my cross-country run, which is about 4 km around the barracks, was above average. Such results were often cause for my superiors to suppose that I would make a fool of them in the shorter, fast-paced events. To be honest, I must admit that I too poked fun and accentuated my inability rather a lot. I do not know why, and that may well be food for psychologists. When we had field exercises and were picked up at the end of the day by 4- or 6-tonners, I could not climb into the vehicles. My mates, one and all, had to hoist me inside. I did find it peculiar, but that is just how it was.

In the military I also participated in the Four-Day March of Nijmegen, 40 km/day, in full pack with two paving bricks in the ammunition pouches to simulate ammunition and to attain the pack weight. I completed the four-day march without any problem, which indeed seemed to be another indication that I feigned some inabilities.

In this period, too, I really had no idea that anything was the matter.

### **Age 20 - 30**

After my term of service I was married and became the father of three children, two boys and one girl, and after one year of marriage we moved to Terneuzen. There I got a job in the laboratory of a chemical company.

Through the staff association I participated in sports, in the beginning volleyball and table tennis. In fact, I enjoyed playing sports quite a lot. In volleyball, however, it turned out that I could not make two motions at the same time. I could smash, but I had to remain standing with both feet on the ground. When I jumped, I could not make any other forceful motion. I found that peculiar at the time, too. Later I went in for football with my department, and the first time everyone agreed that we were all in poor shape and that we must have a rematch once we were in better shape. I then ran about 5 km four evenings a week. I did that for two months, I think, and the striking thing was that I did not improve a bit, and I had to slow down and breathe for a moment, bend/stretch, etc. at practically the same place every time.

Afterwards I went for a few years to a gym club for adult men (anti-belly club), where I could clearly see that I was doing considerably worse than the others. The frustrating thing was that most men performed poorly in the beginning and then gradually benefited from the exercise, while my performance continually diminished despite all the exercise. I even began to exercise fanatically, without result.

Meanwhile, my family developed further, and I soon saw that my children were not at all like their father in terms of sports performance. That was reassuring to me.

In this period I found that I achieved considerably less than others, even with our physical conditions taken into account, but I was not yet worried about it.

### **Age 30 - 40**

In 1972 I read that exposure to chlorinated hydrocarbons could cause liver damage. After contact with the medical service it was decided that all employees would be tested for such exposure.

To my surprise, I was called in to the company doctor because he wanted to see me. It so happened that my blood status was the same as that of:

1. someone who had severe jaundice
2. a typical pregnant woman
3. someone who was completely exhausted.

Since I was none of the three, the company doctor assumed that an error had been made in the test (it may have been a fibrin clot), and so it was repeated.

The result was the same again, and thus there had to be something wrong. There were some substances that might indicate liver damage. I was referred to the hospital in Terneuzen. There I underwent a two-week examination that included spinal puncture, kidney examination, liver examination, and blood test. There were no findings other than the deviations in the blood.

In 1973 I was sent to the Academic Hospital in Utrecht for another examination. I did not get a definite answer there, either, and they wanted to perform another heart catheterisation in the vertical position, but I refused it since my experiences there in the hospital were annoying, to say the least. I had hardly any more confidence in the doctors there.

In this period I began to realize that something was possibly wrong with me, but I did not yet worry much. Ultimately I still functioned normally in my work and at home, and only sports were going poorly, though I still liked to participate in them.

### **Age 40 - 50**

In 1983 I went to the family doctor because I found that my locomotion had begun to be very troublesome. Through the neurologist I finally ended up in Nijmegen, where the diagnosis was made and I was informed that nothing more could be done about it. Thus at that moment I knew that I had a muscular disease, and I became a member of the VSN [Dutch Muscular Diseases Association].

The first annual meeting of the VSN gave my wife an immediate shock, and she was thoroughly stricken. For me it was just the opposite, I saw fellow sufferers, and I felt a little bit normal again. A number of days later the positive feeling turned into a sad feeling, probably because I realized just then that what had happened to me was serious.

A year later my wife had a brain operation, which was just as alarming. Our reaction was: Is everything going to happen to us now?

My locomotion began to worsen gradually and continually. The frustrating thing was that as soon as an activity dropped out I just knew that it would never come back. The operation on my wife was successful, and so that was a relief. Meanwhile, the children were going to university in the urban agglomeration, and the two of us stayed in Terneuzen.

We had a home with three storeys, and we anticipated that it might become more difficult in the future, and we decided to move to a low-rise building (1988). For my job I had to travel by air sometimes, and it became increasingly difficult to step into the transfer buses at the airport and to board the airplane. Because of my height of 1.86 m I had to bend slightly to come aboard, but as I bend I go further forward, and so one time I fell into the arms of a stewardess. Despite the fine and friendly appearance of this lady, this was no cause for me to be happy, too.

In this period we had difficulty in our relationship with each other, especially since we tried to spare each other. Neither one of us had a good idea of how we should deal with the disease. The curious thing was that I myself did not consider it as serious as my wife saw it.

### **Age 50 - 60**

I no longer went to work on a bicycle but on a Spartamet [motorised bicycle] (1992). As a result I did not feel as free as I had otherwise. I noticed some resistance to overtaking other cyclists since I had earlier experienced that the passed cyclists then went on to make annoying comments, such as: You are still young but too lazy to just pedal. You learn to deal with this too, of course, but all the same.

In 1992 I was given a different position in the company, and I had physically nothing more to do with the laboratory. I had been permanent manager of the laboratory and product steward (who checked the products to see whether they met the laboratory requirements) and was now full-time product steward having contact with the customers. This move was partly due to my difficult locomotion in the laboratory. The new position was entirely an office job. It suited me (and the others) quite well.

In 1994 there was a restructuring of our company, and since I was the oldest in the building where 150 people worked, I received an offer to retire early. At the time some 100 persons retired at 55 years of age. At first I did not really want to do it, but in retrospect it actually turned out quite well.

When driving a car I noticed that I began to have trouble with a drooping eyelid. That lid drooped exactly halfway over the lens, and always when I was tired. In 1996 an eyelid correction was performed.

Wishing to know whether I used my well-functioning muscles as efficiently as possible, I went to the rehabilitation doctor in 1998. That visit resulted in the purchase of a stand-up chair and the instruction that I must inform the CBR [Central Office for Motor Vehicle Driver Testing] that I had to drive a car with an automatic transmission. I myself had already got one since I found it easier than a car with a standard transmission. As of that moment I therefore had to apply for a new driver's license and (once again) be tested. That driver's license was valid for 3 years, and after that it began over again.

So with the two of us in the house there was at times quite some tension about dealing with the disease. As a patient I was inclined to do everything myself as much as possible, and my wife could

not bear that I had to take such pains to manage some simple thing. It was so much easier if she just did it herself. I did not find that acceptable, as I was a bit proud of my own results.

The opposite was also true sometimes, when I misjudged an activity and could not manage it, while my wife knew that I wanted to do as much as possible myself and therefore did not help. I did not find that very amusing. Thus it really never seemed to be good.

In this period came the realisation that there was no return, and your future looked utterly dark. We were both full of sorrow, and like old people we began to make more and more use of the pleasant memories that we had had.

### **Age 60 - present**

Walking was no longer easy for me when I was on uneven ground. Through the rehabilitation I was advised to get a walker and to apply for a handicapped parking card. After (more) tests I obtained such a card in 2000. Remarkably, I found it in myself to do my very best at these tests in order to make as good an appearance as possible, which was actually to my disadvantage. It was no longer possible for me to stand up from the toilet. At the beginning of 2000 a toilet lift became necessary. This was installed in the upstairs bathroom.

After awhile the eyelid was corrected for the second time.

Also the Spartamet was used less and less and at some point no more at all. I could no longer use it.

At some point I could not even climb stairs, and we have been sleeping downstairs. For that reason an alteration was made through the WVG [Services for the Disabled Act]. The small downstairs bathroom was made larger, and the toilet lift was installed in it (late 2000).

In 2001 I went to rehabilitation again because I was doing really quite poorly. There they clearly agreed with me; for they saw to it that I was provided with a push wheelchair and a *scootmobiel* [powered wheelchair] (Booster brand) and advised me to buy a sliding sheet. I could just barely turn over in bed, you see.

At home, however, it was increasingly difficult to do activities, especially to use the bottom drawers of a chest. Standing up from a chair became impossible, too, and a stand-up easy-chair and *trippel* chair [work chair developed for disabled people] were now necessary. Outside I still move about but only with the wheelchair, whether motorised or not. At present we can no longer sleep at the children's because all the facilities are upstairs.

We now have 7 grandchildren, which means that our children could very well ask us to baby-sit or to do other small jobs. They all live two hours away by car, however, and so we would really have to sleep there. We did that a few times in a hotel in the neighborhood, but that too became increasingly difficult. When the children come to us with the grandchildren, the toddlers rush at Grandma and Grandpa, but Grandpa must then sit down quickly or else he would be run over.

Whenever Grandpa lies on the ground he must be helped up again with much manpower. I can hug the children only if someone else holds the child directly in front of my nose.

Nowadays with all the accessories at home I can keep busy quite independently. I still do some volunteer work.

During my life I have always done something besides my job, such as: treasurer of the parents' committee, Intercaritas chairman, badminton coordinator for holiday makers, treasurer of the mixed choir, chairman of the Voluntary Home Care Network, secretary of the Voluntary Home Help Reporting Centre, parish membership records.

Now I only do the Voluntary Home Help Reporting Centre, where I have telephone service once every two weeks and am secondary secretary. That mainly involves work at the telephone and the computer. I attend the meetings on the wheelchair.

I still keep the church membership records up to date, and that too is mainly work at the computer and attendance at meetings a number of times. In addition, I sit in the church choir, which involves a weekly choir rehearsal plus singing in the church a number of times. I go there on an electric wheelchair, and my place is at the back, and I can raise the chair so that I can see the conductor. It is not possible for me to sing in all the churches because sometimes the choir stands on a platform, and I cannot go up there. I go along on the annual outing with the choir to another church, but with my own transport since it is impossible for me to step into the bus. I have stopped a number of these activities because of my disease. I have found that I cannot fully function. For example, when you yourself can no longer play badminton, you do not want to be coordinator anymore.

I have played a great deal of sports, and again and again I have had to switch over to another sport that I could still practice. The sports were football, volleyball, table tennis, and badminton, and at present my wife and I play bridge with the seniors of the staff association and of the parents' health foundation in Terneuzen.

In general I cannot use public transport. Stepping into the bus, standing up in the bus or train, and climbing station steps are all impossibilities.

I cannot even use a normal taxi because I cannot get out.

In February 2005 my wife underwent a heart operation, and she now has a pacemaker. That gives us both an unpleasant feeling because each reduction in the activities of the Pompe patient means an increase in the activities of the other.

In this period we have also requested that once every two weeks a helper from Home Care do the difficult work, such as mopping the floor, washing windows, changing bed sheets, etc.

Up till now my personal attitude toward the disease has been to ignore as much as possible and make the most use of what I still can.

It has the disadvantage that many do not see, and thus do not know, how little I can still do, and therefore I am asked to do all kinds of things that are not possible for me. The advantage is that I am still usually approached as a normal person, and I feel pleasantly included.

My feelings about the future are more difficult to explain. On the one hand, I do not think about my future in much detail since that would be depressing, and on the other hand you have to think about it in order to apply for aid in time. My wife is much more precise in the advance planning and worries about it sooner. Understandably, of course, as she is the one who provides care, and it only increases. Logically, she does not have a cheerful perspective.

With the aids I can manage fairly well, and I am happy about that, but the processing of the applications has caused much distress and anger. To me it is incomprehensible that with a disease such as I have you must explain what exactly is the matter all over again ever time. Even in the same institution, e.g. the Municipal Authority, you have to run through the same story every time. The first few times I had a terrible amount of difficulty there, especially since you have to tell the story to a counter clerical worker who has no medical training. I had the feeling that I was there to satisfy other people's curiosity.

The feeling that now dominates my situation is not that I mind the wheelchair and other aids but that I would be very glad to be able to stand up from a chair or to get out of my push wheelchair. My wife must always lift me up, and that involves a motion similar to a very intimate embrace. For me that is not bad in principle, but to do it before the eyes of strangers is another story. Thus you can never sit down anywhere, e.g. on terraces, at other people's homes, in the bus, in the train, etc., without having to be lifted up from the chair. Now I just happen to be a tall man, and so it is all the more a spectacle to watch. I get out of bed with the help of the *trippel* chair. This is placed next to the bed with the seat and back and arm rest at the proper height (which requires considerable precision), and it will work just as long as it lasts.

I can still clean myself after going to the toilet, and I am quite happy about that, just as I am about the fact that I have a very regular bowel movement. Every morning after breakfast is my time. Thus for the rest of the day I do not have to be concerned with having to go.

My car is adapted. It began with an automatic transmission and then a special driver's license that allows me to drive only a car with an automatic transmission. Now I have a hand brake on the steering wheel for emergency stopping because I am afraid that my leg is not quick enough to react in all situations. For example, when I make a right turn, my leg swings to the left and cannot return to the pedals in time. My feet work well, and so when my heel is on the ground I can quickly turn around anywhere on my foot with the heel as a point of support.

Stepping out of the car became the big problem. Having purchased a higher car (Renault Scénic), I later had to have an aid added. A chairlift that raises the seat and turns it a little bit outward has been built in. For the time being I can get out again.

The latest developments of my disease, at least to my thinking, are the swallowing movements and chewing. During meals my chewing, which was never rapid to begin with, is becoming slower and slower.

Swallowing, in itself, still goes well, but mucous remains at the back of my throat, and it is difficult for me to get off. Also whenever there is a crumb on the back of my tongue I cannot get it off, and I begin to cough and sneeze, and my eyes water. According to the speech therapist at rehabilitation, the rear section of my tongue does not function well. I am also beginning to notice that with talking. For that reason I am beginning to withdraw from functions in which you have to speak at a meeting. Sometimes when I begin to talk a wave of saliva comes all at once, and I reflexively begin to wipe with my hands, and I find it quite annoying.

In bed I use a flannel blanket on the pillow because I sometimes lose a great deal of mucus in the night. Recently I have also noticed that I choke more and more frequently.

Recently I have also noticed a considerable weakening of my arms. Combing my hair is difficult, and drinking from a glass in a standing position (when brushing my teeth) requires two hands. Because I sit a lot, my bottom is beginning to feel more and more as though it were asleep.

Summary of feelings:

Gratitude that there are so many aids that I can use and anxiety about where all this is heading.

There will come a time when I will no longer be able to use even the aids.

Gratitude that we have three fine children and 7 nice grandchildren, but sorrow because I cannot do with them what I would like to do.

In general I can give only a poor description of my own feelings since a dominant quality of mine is to ignore all the negative things and go on to positive things. The idea is that when you dwell on pain or disease, you only make it worse. The problem is, of course, that there are more and more negative experiences and fewer and fewer positive ones. I once described my feeling as if you lived in a room that was always shrinking. At first you can still jump high, then you can just stand erect, next you can only stoop, after that you are lying down, and what follows then?



My family and I (left)



Enjoying nature together



Together on the path