**A NARRATIVE OF POMPE'S DISEASE**

I noticed the first abnormality at the age of 36. Had I not been medically qualified, I doubt that I would have been aware of the problem. After emerging from a bath, I noticed that the left side of my lower abdomen seemed more protuberant than the right and my belly button was not central. It did not take long for me to determine that my left lower rectus abdominis muscle was paralysed.

I arranged a consultation with a neurological colleague. He said that the only thing that could cause an isolated paralysis like this was poliomyelitis, and that at some time, I must have suffered an undiagnosed attack. Surprised, but somewhat reassured, I took no further action.

Nothing further happened until I was 40, when I noticed severe wasting of my right pectoralis major muscle. There was little to show in the way of weakness and I was still playing tennis and squash. Again, a neurological consultation led to a very unsatisfactory diagnosis; congenital absence of my right pectoralis major! I was quite unable to accept that I had not noticed it for forty years. Me, with my narcissistic pride in the body beautiful.

I worried about this for some months and then I noticed wasting of my latissimus dorsi and trapezii on my back and neck, accompanied by some restriction in lifting my arms above my head. I had also noticed episodes of fasciculation (twitching of small areas of muscle) in my arms and legs, which is a common symptom of motor neurone disease. Yet another neurological consultation was a farce. As soon as I mentioned that I was worried about the possibility of motor neurone disease, my neurologist jumped to the conclusion that I was severely anxious and depressed and suggested referral to a psychiatrist. By now, all faith in neurologists had evaporated, so I did arrange to see a levelheaded psychiatric colleague. He agreed with me that my problem was muscular, not psychiatric and arranged a new neurological opinion at another teaching hospital.

By the time that I saw the new neurologist, my left shoulder had become painful. He was a thorough and knowledgeable doctor, and he agreed about the wasting and weakness of my abdomen and shoulder girdle, but he seemed uncertain about a diagnosis. Finally, he homed in on the recent shoulder pain and said that it could be 'neuralgic amyotrophy', a condition that usually follows an injection or vaccination. He suggested a consultation with a rheumatologist about the painful shoulder and said that if he was correct in his diagnosis, the muscle weakness could improve over time. His rheumatological colleague was delightful. He said that he had no clue as to the diagnosis but felt that a steroid injection into my shoulder could help the pain. Within a week of the injection the pain had gone.

By now, my faith in neurologists had plumbed the depths. Having consulted countless texts, it was clear to me that none of the diagnoses fitted the facts. As luck would have it, I was due to be an external examiner in medicine at a medical school where a world authority on muscle diseases worked. I wrote and asked if he could see me while I was examining there, and he agreed. Indeed, I got double value as he asked another muscle expert to sit in on the consultation.

The history taking and examination were detailed and thorough. They both agreed that I had 'benign spinal muscular atrophy'. This is neurological jargon for motor neurone disease with an abnormally slow deterioration. In other words, they confirmed my own self-diagnosis with the addition of the word benign. They also agreed that I was likely to reach my seventh decade, although, as they put it, ''you could be in need of a wheelchair''. I was told that the diagnosis could be made more certain by having biopsies and electrical studies of muscles and nerves. As this would be purely academic, in that no treatment was available, I opted not to pursue the matter further.

For the next twenty years, I followed my medical career without seeing another neurologist. Progression was exceedingly slow. In my mid-forties, I abandoned squash and tennis. By my mid-fifties, I began to use a stick on long country walks but my disabilities were small. In my mid-sixties, my wife could see that my walking and balance were obviously deteriorating and urged me to seek a further neurological opinion. One does not tangle with she who must be obeyed, so I made an appointment with a young neurological colleague. He agreed with the diagnosis, but said that he was not an expert on muscular problems and offered to refer me to someone who was. Here I made a big mistake. We discussed what such a referral would achieve other than a more precise diagnosis and I decided not to bother. I merely arranged to see my colleague again in six months.

Before the six months was up, I noticed increasing daytime sleepiness, causing problems working and driving. Again, my wife persuaded me to see my colleague. On hearing of the sleepiness and noticing my paradoxical breathing (a sign of a paralysed diaphragm), his eyes lit up and in a eureka like moment, he said ‘‘you have acid maltase deficiency!’’ He arranged for a blood test for vacuolated lymphocytes and also arranged a sleep study for the following week. The blood test was positive but I never made the sleep study. On the following Saturday morning, my wife was unable to awaken me. I was in severe respiratory failure. Fortunately, I was immediately transferred into a medical intensive care unit.

I had a very stormy three to four weeks and was given a poor prognosis. I finally came out of my coma in the fourth week, with a tracheostomy and a naso-gastric feeding tube. I spent a further six weeks in the unit before being discharged home using a ventilator at night and a suction machine to clear the tracheostomy when necessary. Those who looked after me in intensive care had previous experience with respiratory failure in Pompe’s patients. They reassured me that, over time, I would return to my former level of activity, and they proved correct. It took at least six months to get back to being a moderately active sixty five year old whose major problems were a tracheostomy and the need for nocturnal ventilation.

I was advised to contact and join the Association for Glycogen Storage Diseases (UK). This was wise advice indeed. After attending a couple of their annual conferences, I realised that the majority of late onset Pompe sufferers did not have tracheostomies, even if they were much more physically disabled than I was. Armed with this information, I set about convincing my carers that closing the tracheostomy would be a good idea, and so it proved. It was closed without problems and since then I have been ventilated via a nasal mask. It was only then that I realised what a major nuisance tracheostomies are. Using a suction machine to clear it out is a deeply unpleasant experience.

The eight years since respiratory failure have been years of rude health. Respiratory function has not deteriorated. I use a respirator at night or when lying flat. I do not need it during the day. Muscle strength has continued to ebb very slowly. I can still walk with one stick, albeit at snails pace. On the flat and with an even surface, I can manage between half and one kilometre before needing to rest. On normal stairs I can cope with one or two flights, providing there is a handrail. Going uphill or downhill is very difficult as is walking on rough ground. Weakness of the trunk and hip girdle mean that I am very unstable walking and the slightest trip causes me to fall, often with no chance to save myself. So far, I have not sustained any serious injury other than very unattractive black eyes and a seriously haemorrhaging nose. Falling is one of my greatest concerns, and has led me to abandon using public transport such as the tube. I am also at pains to avoid crowds.

One serious lesson I have learned is not to use a strange bath without much thought! Always inspect it very closely to make sure one can get out. Calling on one's host or hotel staff to help one get out can be quite embarrassing.

Progression has been amazingly slow. I now expect that some intercurrent illness will get me rather than the Pompe's itself. I have to admit that the idea of an epidemic of 'flu or bird 'flu is quite scary. I have also become aware that exhortations to exercise and keep fit are almost impossible to fulfil when one is disabled. The options available like exercise bikes are mind numbingly boring. By far the most important advice to a late onset pompe patient is, make sure you have a devoted wife, husband, partner or parents. In this I am an exceedingly fortunate man.

Malcolm