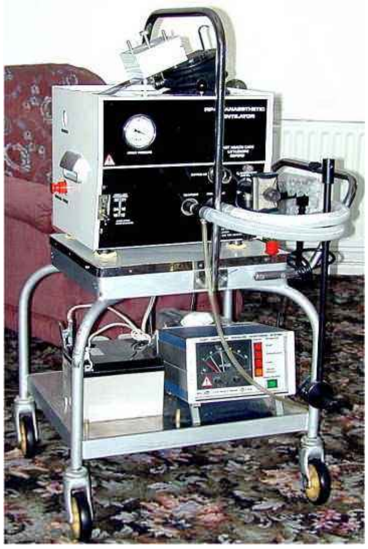
Johns Story about Pompe and treatment with enzyme replacement therapy

September 2007 (age 64)

I thought I would let you know how John (aged 64) is doing after seventeen infusions. As you know for the first 26 years of John's illness we had no input from anyone. But for the last 3 years we have had two nurses on our side and thankfully they do their best to help in anyway they can. Social services and Rotherham District Hospital (RDH) are lacking in a lot of ways, but now John is under The Hope Hospital at Salford, Manchester things are so much different. At Hope he has 3 physicians looking after him. They are all so very good and show such a lot of interest in John which he has never had before.



The Radcliffe ventilator John has been using for years.

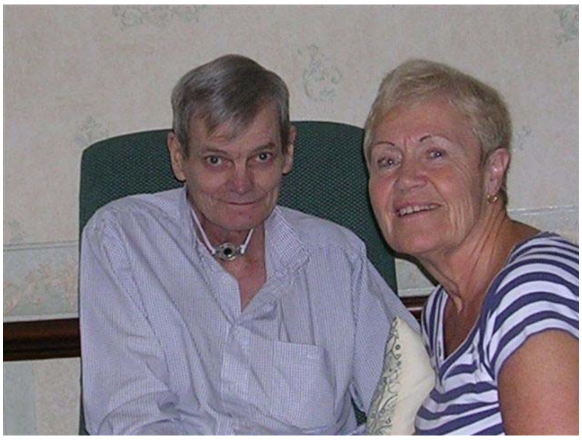
When John first started his infusions he was in a very bad way. Two years before the men who work in the BME department at RDH had said that the two old Radcliffe ventilators that John had been on for years were obsolete and they could not get any more spare parts for them. So they tried him on four different types of machines (at home) with me in charge after half an hour tuition and a CD to learn from. John could not get used to any of the machines as they were all so different to the old east Radcliffe, which you probably know had bellows and weights on the outside of the machine which just clanged up and down gushing the air into John's lungs at a pretty fast rate. Nobody had any idea of how much ventilation John had had over the 26 years as nobody had ever seen him to check on his ventilation or anything else; the outcome was that John's lung capacity is twice the normal size because of the over ventilation. With the new machine delivering the ventilation so differently from his old machines, John has not slept properly since. He is lucky if he gets a couple of 10 minutes every night. He has had sleep studies done at St. Thomas' Hospital in London and they did not know why he was not sleeping. I am pleased to say that they all agree with me that it was the changeover of the machine that started it off. It had to be that as it happened on the first night of the changeover.

As a consequence of this change of sleep pattern John deteriorated rapidly. He became very weak, went off his food, and became very low in spirit. He could not help himself much at all. He had a chest infection that led to Pneumonia and that took its toll. He went really downhill. He then had a fall which damaged his knee and he became really very dependant for everything. Gradually his knee got back to normal but he was so very weak that he never went out of the house for two years because he could not help me to get him out of the wheelchair as he did before, and to get him in the car was impossible.

At this point I found out about the new LSD expert centre in Manchester and got a referral to see two physicians. They did muscle biopsy and skin tests on John to confirm that he had Pompe Disease, but one of them said as soon as he saw John he knew he had Pompe's. The next time I saw this physician was at the AGSD (UK) conference at Bradford. Our Community Nurse told him how ill John was and he said that John's muscle biopsy was the worst he had seen. John and I presumed that with this news about his muscles and his age it was more than likely that he would not be getting the treatment. However the following week we had an appointment at the Pompe clinic at the Hope Hospital.

I know that both physicians were shocked to see how much John had deteriorated. He had lost weight, he could not sit up properly without support, his voice was very weak and he could not hold a conversation longer than a couple of words. He also had to support his neck with his right arm as his muscles were so weak. John told one of the physicians that this was the worst he had felt through the 28 years of this illness, and that he felt very low because he thought that he could not halt this deterioration and that he was just going to get worse. The physician was brilliant. He told John that they were going to help him as much as they could. They said they were going to admit him to Hope Hospital for a week to do several different tests and if they were all satisfactory they would give him his first infusion on the Tuesday of the week he was staying there. We just stared at him in disbelief. As I said earlier we thought we had little chance of him getting the treatment. We were elated.

And now, here we are seventeen infusions on and he is like a new man. Before he started the infusions he could only manage 6 hours out of every 24 off the ventilation. He was on 22 breaths a minute and we have just got him down to 16 breaths a minute and he is now off the ventilator 12 hours out of every 24, which is fantastic. He is back to walking (or waddling as the doctors call it) around the house, his eating has improved, he has put on weight and best of all, with a lot of effort from both of us, I can take him out in the car. I can manage to slide him into the car and when he gets out I lift while he pushes down and I rest him on the side of the car and he climbs up the door (it's not as hard as it sounds). The only problem we have got at the moment is when I take him in the car he has to stay in it till we come back home as we have not mastered the wheelchair bit. I can get him out of the car into the wheelchair but as yet I cannot get him out of the wheelchair into the car. But we are working on it and I am sure we will get there.

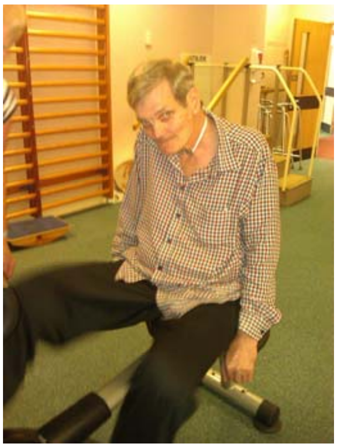


John and Val

Three weeks ago we went for John's six months mini-assessment and the physicians were very pleased with John's progress. One of the physicians said that 12 hours off the machine was amazing. He also said that John's voice was a lot stronger and that he was pleased he could have longer conversations. He was also surprised that John was not having to support his head had like he had been doing. The neurologist did some muscle strength tests and said both of John's arm and leg muscles were stronger. He had John sit on the bed with his feet pointing upwards and said he was going to push John's foot down on to the bed. I was standing behind the physician when he was trying to do this and he really was shaking with the effort. He then stood up with a smile on his face and said: "you all know I am a chap but I could not push it down. His leg there is normal". The muscles John has always been able to use are getting stronger all the time. The ones he lost the use of due to lack of physiotherapy and other reasons are not showing much improvement at the moment but we are not going to expect too much all at once. We are doing exercises on these muscles ourselves to try and work with the treatment. As far as we are concerned what John has achieved up to now is a miracle in itself and I am so pleased and proud that the physicians say that John is their star patient. He pushes himself and does as much as he can to help himself and I know if anyone will benefit from this treatment it will be John. The change in him is remarkable. He is so much happier and wanting to try to do so much more than he has been able to for a long time.

July 2011 (age 68):

His muscles got stronger, his walking got better and we got his breaths on the ventilator down to 12 breaths a minute and 14 hours a day off the ventilator. We managed to go on holidays, something we had not been able to do for a long time and generally get out and about. Things were fine until January 2010 when, due to neglect of equipment by the social services, John had a very bad fall. He hurt his back and the top of his head was burst open. He ended up in intensive care and the high dependency unit for 8 weeks where he was critically ill, and then in a rehabilitation centre for three months. The day he came home from there he started with nova gastric virus (other people on the ward had got it) and he was ill for five days . He could not eat a thing as it went straight through him. He looked so ill. He had lost two and a half stone in weight. his arms and legs were scrawny and worse still he was ventilated 24/7. He could not walk and he could hardly sit upright in a chair. Nobody thought he would ever get back to how he was. But I am very very pleased to say that he has now put the weight back on he has got muscles where he did not have them before and he is going from strength to strength. As soon as we could we started him back at his physio sessions and they have paid dividends. His consultant Dr. Waldeck gave us the wonderful news that John`s enlargement of his heart has now gone. He has a marvellous physiotherapist (John Brinkley) who has worked so hard to find out as he could about Pompe disease and it shows with the wonders he has worked with John. John can work on the exercise bike on level 8 for about nine minutes and he has now started walking again with the aid of a frame or holding on to my arms. We cannot believe the progress he has made. He truly is an inspiration to everyone. He has worked so hard to overcome everything that is thrown at him and I am so very very proud of him. We are confident he will be walking on his own again in the near future and that I will be able to get him out in the car again.



John on the exercise bike

Val and John, United Kingdom