Jamie 6/12/89

Jamie was born 7 weeks early at 33 weeks gestation, weighing in at 6 pounds and 14 ounces. During my pregnancy I had a loss of blood at 20 weeks gestation, preceded by a fairly severe pain from a fibroid or two. From then on everything settled until Jamie arrived after a 6-hour labour and normal delivery.

Up until the age of 6 months everything seemed to be progressing well despite the fact Jamie remained a floppy baby, but this was always put down to his being very premature. Jamie started to get upper respiratory infections sometimes affecting his chest. He also started to develop obstruction of his airway when put down in his cot to sleep; this resulted in him waking frequently usually with vomiting. I noted when I observed him his tongue flopped back obstructing his airway.

Jamie was referred to a paediatrician at 14 months by which time I had become increasingly concerned, as he seemed to have developed sleep apnoea and was very unstable when walking; his gait was unusual. From the initial referral he saw three other consultants: 1.) Ear Nose and Throat 2.) Orthopaedic and 3.) A Community Paediatrician who looked into his development of motor skills and his hypotonia. These referrals went around in a loop for a further 9 months to no avail, Jamie still had not got a diagnosis; all the consultants were baffled by his symptoms. Sadly Jamie experienced a respiratory arrest in January 1992 at the age of 2 years and one month at 8 o'clock one morning. It was only by chance that I found him, just as he had collapsed and his heart stopped beating. I resuscitated Jamie in at home; he was then admitted to hospital as an emergency, he was unconscious, and convulsing. He remained on intensive care for 3 days intubated, ventilated and sedated. 7 days later he was transferred to Great Ormond Street for a second opinion. We had refused a tracheotomy procedure on Jamie at this stage and demanded that we should be seen in a specialist hospital. The anaesthetist was very understanding and supported us with this decision.

At Great Ormond Street Hospital Jamie was eventually taken to theatre and had his tonsils and adenoids removed, this procedure seemed to rectify the apnoeas and the muscle biopsy taken at the same time revealed Pompe disease as the cause of Jamie's problems.

Between the age of two and the present time Jamie reached many developmental milestones on schedule. Throughout his early days at nursery school I attended more than other mothers to help Jamie so that he could lead a normal life. If he wanted to climb I was there too, climbing along side him. At school he was in a very small class, with an assistant in the playground to keep a close eye on him whilst he was at play. He did remarkably well despite his physical limitations. His ability to persevere with most tasks was helped by his good nature and acceptance of his own limited strength. He was able to learn and was sociable. I have always kept very close contact with schools and other families who cared for him in my absence. I can't deny that his mobility has been a worry and still is; despite the fact he manages very well and is now young man aged 15.

From birth Jamie has always found it hard to settle and sleep, night after night we had problems with him getting too hot and often the only way to settle him was the

comfort of being with us. It was a major problem. His breathing was frightening at times. Eventually in 2002 at the age of 12yrs he had a study performed on his sleep pattern. He had started to develop morning headaches and felt sluggish and had difficulty concentrating. It was discovered he had sleep apnoeas whilst in the deep part of sleep/REM (rapid eye movement). From this moment on he started to use a machine called a V-Pap, providing assisted ventilation at night for about 8 to 10 hours daily.

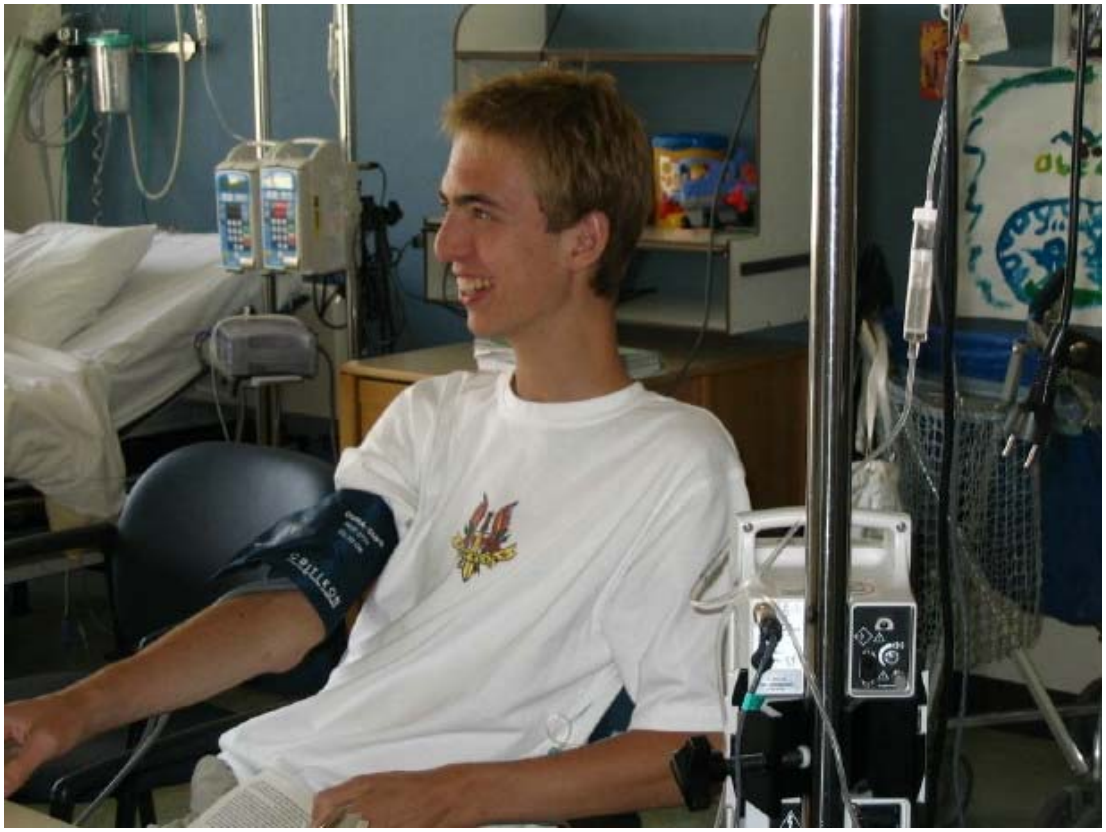
As Jamie grew taller, rapidly during puberty, it became apparent that his spine was developing a scoliosis (sideways curvature). By the age of 13yrs the degree of curvature was so severe it was recommended that a posterior spinal fusion be performed to straighten his spine in April 2004. The lumbar curve was at this point approx. 60 degrees and the thoracic spin approx. 30 degrees. Jamie gained 2 ½ inches in height after the operation. Before this time a spinal brace had been not only very uncomfortable but also ineffective for one year.

Jamie recovered from this operation and returned to school after eight weeks. He was encouraged to walk regularly to regain strength. Three months after the operation he was allowed to swim and at six months start playing a little gentle tennis (the sport he loves). Now, after eighteen months, he is very mobile and only suffers backache after prolonged exercise.

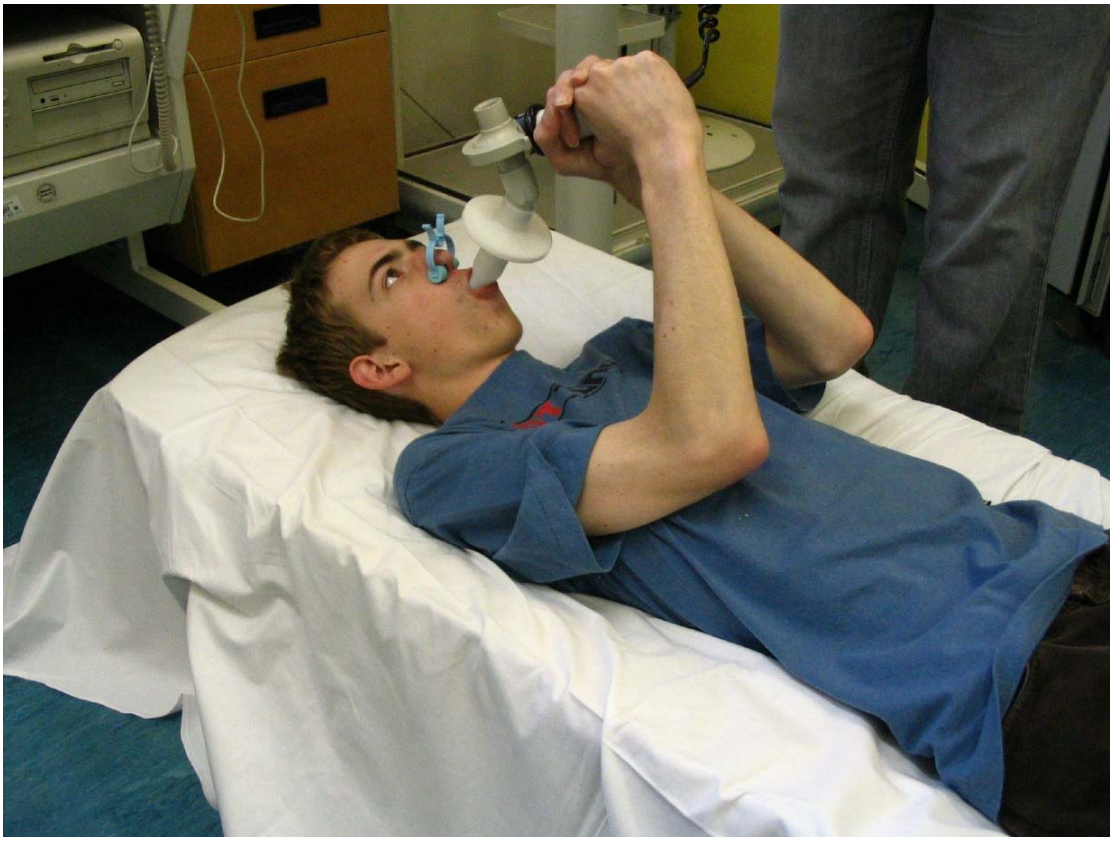
Jamie is very slim at 6 feet 1½ inches in height. Since the age of diagnosis we have encouraged a diet biased towards protein and discouraging high carbohydrate content. There have been periods when we have used high protein supplements that in general have been unpleasant to the taste. He also has had 12 grams of L-Alanine per day as a supplement to his diet since the age of 8 years. He has a good appetite and has no problems with chewing or swallowing.

To conclude, Jamie is at present aged 15 years and 11 months. He has always attended state schools in the UK, integrating well and participating in most activities and academic studies. Nine months ago Jamie was fortunate to be enrolled onto a clinical trial of Myozyme, Enzyme Replacement Therapy for Pompe Disease. He is tolerates the therapy well and appears to be responding positively, we are very hopeful that the progression of the disease will halt, or better still reverse, in the near future.

By
Barbara (Parent)



Jamie getting his infusion



Jamie doing a lung function test

Jamie drawing a painting

Jamie on a boat on holiday