Jayne’s story

The fourth in a family of eight children, my sister, Jayne, was a gentle child who loved sport but for some mysterious reason, always had trouble running and keeping up with her team mates. Despite this, she took what we all thought was just her particular inability to run well with good humour and enjoyed the camaraderie that comes with being a member of a basketball team over several seasons.

As someone with enormous compassion for others and a selfless capacity that sometimes worked against her own best interests, it was no surprise that Jayne chose nursing as the profession she would enter on leaving school.

From the outset, Jayne brought a sensitivity and concern to her work that never wavered, no matter how long the shift or how intense the work. Universally respected and liked by her colleagues, Jayne was renowned for her willingness to go the extra mile. One of her nursing workmates relayed the story of Jayne’s concern for an elderly lady with dementia who was distressed because she had to wear a hospital gown while an in patient in the hospital. Without a family to bring her own clothes to this elderly lady, Jayne went off in her lunch hour and bought 2 pretty night dresses for this patient to wear that soothed the latter’s concerns and turned this lady’s stay in hospital from one of distress to one that was happier and more productive for all concerned. Jayne neither expected nor received any recognition or recompense for these actions.

Over the course of her career, Jayne’s professional capacities and expertise were recognised by senior surgeons who requested Jayne’s presence in theatre and invited her to assist in pioneering surgeries that were being trialled.

As well as succeeding in her chosen profession, Jayne had always wanted to marry and raise a family and at 24 years of age Jayne married her husband Corrie. One year later she was overjoyed when she gave birth to her first son, Dan and 18 months after that was thrilled when her second son, Marc, was born.

Jayne believed her life was complete. She had a husband she loved and sons that she couldn’t wait to see grow and develop. She had a highly successful career and a job that she intended to return to when family commitments allowed.

Two months after Marc’s birth, however, Jayne began to notice that her legs would suddenly and inexplicably give way. She started experiencing a growing number of falls and because she was now caring for a 2 month old baby, was extremely worried that she would drop and harm Marc in one of these sudden falls.

Jayne went to see her local GP who could find nothing overtly wrong, though there did seem to be some weakness in her neck. This was the second inexplicable result she had experienced, the first coming from a liver biopsy that showed similarly inexplicable abnormalities when she was 22 years old.

Jayne was surprised that nothing more obvious could explain what she was experiencing and even more surprised when her GP referred her to a neurologist for further investigation. The neurologist performed a test that showed muscle abnormalities and then referred Jayne to a surgeon who performed a muscle biopsy.

Jayne’s marriage was just three years old and her sons were only 18 months and 8 weeks of age when she was told she had Glycogen Storage Disease Type 2.

I can clearly remember this time as my parents and I were looking after Jayne’s boys while she stayed in hospital for 2 days to undergo this surgery. I remember how Dan screamed and refused to leave his mother when we left the hospital and how my father drove us home while in the back seat, I tried to restrain a despairing and frightened child who couldn’t understand why he couldn’t stay with his mother.

I also remember the day we heard that Jayne had Pompe's Disease and how stunned we all were. For this to happen to someone as gentle and loving as Jayne seemed the cruellest thing of all. That it had happened to a young mother at the start of her life was bewildering and our bemusement quickly turned to questioning the doctors about the accuracy of the diagnosis before finally descending into anger and despair.

Some of my family simply couldn't contemplate what this meant. We had already lost a brother and a sister to the ravages of Cystic Fibrosis and it seemed extraordinary that we were once again facing a disease for which the medical profession had no answer. My way of coping was to find out everything I could about this strange and unheard of disease. I was determined that I would find a way of helping my sister cope with what she faced and that an answer would be found to heal her.

Jayne's husband found it particularly hard to comprehend this radical change in his family's circumstances. Corrie had never experienced illness and when he had confronted it in others, had responded by removing himself from contact with the person concerned and pretending nothing was wrong. He found it difficult to enter a hospital and viewed doctors as people who really had little to offer, believing that people recovered from ill health by themselves and were the better for it.

This attitude ultimately destroyed his marriage and helped him blame Jayne for having this rare disease, a position he maintains to this day.

Over the next several years, Jayne's condition gradually deteriorated. Like many others with neurodegenerative diseases, she eventually had to accept using a walking stick and I remember how we tried to ease the fact that her condition was slowly worsening by decorating her stick and insisting it 'make a fashion statement'. We aimed to have one for every outfit and start a new trend amongst stick users!

But unfortunately this wasn't the end and by the time Jayne progressed to using crutches to help her walk, the efforts to make these aids into fashion statements seemed frivolous and ultimately pointless. We were all being forced to confront the awful truth that Jayne was getting worse and there was nothing we could do about it.

Over the intervening months and years, Jayne's legs continued to give way and she suffered a broken coccyx, fractured hand and arm, broken rib, broken nose and many, many bruises. A particularly nasty fall resulting in a fractured pelvis and broken femur, was also the fall that put Jayne into a wheelchair for good.

With advanced osteoporosis caused by the malabsorption that accompanies Pompe's Disease, she spent 6 weeks in hospital and a further 6 weeks in a rehabilitation facility where, amongst other things, she learned how to transfer using a transfer board, confronted the fact that she would never walk again and began the slow, painful and devastating process of adjusting to life as a person with a disability in a wheelchair.

During the time Jayne was in hospital, another sister and I slept by Jayne's bedside for the first week and then went into the hospital in shifts throughout the day, starting at 6am and leaving at around 9pm. We did this because, despite the best efforts of the nursing staff, they had enormous difficulty understanding that Jayne's muscles were so weakened that she could not comply with many of their instructions and requests. Because of this and in spite our best efforts to prevent it, several situations occurred in which Jayne experienced additional intense pain because the staff thought she was unwilling to try to move. Not only did Jayne endure the additional physical pain this delivered, she was intensely frustrated and depressed by this latest loss of her independence and struggled to come to terms with what this would mean.

This long stay in hospital not only placed Jayne permanently in a wheelchair, it also revealed how far removed from Jayne her husband had become.

In the years since her diagnosis, Corrie had struggled and ultimately failed to come to terms with Jayne's illness and its ramifications. Sadly, this is a situation often confronted by those suffering from chronic and degenerative diseases. Almost from the outset, Corrie refused to acknowledge Jayne's illness and responded to her as if she was in the best of health. This meant that, despite her increasing frailty and dependency, he maintained a view that nothing was wrong and that Jayne was quite fine. In refusing to acknowledge her growing need for support and assistance, Corrie ultimately transferred the responsibility he should have shouldered to his two young sons.

Again, in a situation that is sadly not uncommon, Jayne's two boys became their mother's carers from the moment they were old enough to understand and do so. The boys have never seen this as a problem and have embraced this responsibility with love and a sense of normalcy that has been humbling to see.

But her illness and the responsibility the boys shoulder for their mother's well being has also left Jayne with an enduring sense of guilt, anger and sadness at what she sees as the unnecessary and loathsome burden she has forced on her sons.

With the fact that she would never get out of her wheelchair forcing him to finally acknowledge that Jayne's condition was slowly but irrevocably deteriorating, Corrie walked away from his now highly dependent wife and abandoned his family. He left his 11 and 12½ year old sons to face the almost incomprehensible responsibility of being the sole carers of their mother.

The fact that they are their mother's carers has never been a negative for Jayne's boys. But their father's abandonment has irretrievably damaged their relationship with him and left Jayne's older son deeply and permanently troubled by the added responsibility he felt he had to bear as 'head of the household', forever worrying that he would fail. (I remember one day looking for Jayne's oldest son, Dan, and finding him curled on his bed, silently crying. I asked him what was wrong and he told me he was very sad because he'd had a dream the previous night and in his dream his mum was running and chasing him and he was so happy. All he wanted was for her to walk again.)

The loss of her husband not only devastated Jayne emotionally. At a time when she had never needed his support more, it devastated her financially by forcing her onto a disability pension and the poverty that accompanies this. It also completely destroyed the last vestiges of her remaining health and dramatically increased the rate of her physical deterioration as she shouldered the physical, financial and emotional responsibility for keeping her family intact. In the two years after Corrie left, Jayne's rate of deterioration increased exponentially despite all that we, her family, did to prevent this.

And we did try. My parents moved from the country and settled 2 kilometres from Jayne. Another sister moved from the country to be near and to help. Yet another sister and I tried to buoy Jayne's parenting of her sons in whatever way we could and began spending increasing amounts of time with her, supporting her physically and emotionally.

Long before Corrie's departure, Jayne had made a decision that what remained of her life would be given unreservedly to her beloved boys. With no hope of a treatment to reverse the ravages of Pompe's Disease, Jayne decided to do everything she could with the boys while she still could. With no hope left, she willingly traded her remaining health for whatever involvement she could have, her one wish being to live long enough to see her sons reach maturity and independence.

In this, she had to endure a relentless campaign by her immediate family who tried to preserve her health and keep her safe in the hope that, somewhere soon, a treatment would be developed. She endured all this 'interference' with the same grace and determination that she has always had.

Her weakened diaphragm now meant that Jayne needed a machine to help her to breathe and her worsening physical health meant an electric hospital bed and a sophisticated pressure mattress now entered her home. This was rapidly followed by the advent of an electric hoist – ceiling mounted to maintain efficacy and hide the hospital like atmosphere that now seemed pervasive – and an electric wheelchair with full pressure cushioning on the seat and spinal areas.

By now, Jayne had lost most of the major muscle mass on her body. We were on a constant mission to keep her feeling warm and I began a new relationship with mountaineering shops in the quest for light weight outdoor gear that Jayne could tolerate and that would keep her increasingly skeletal body warm.

The indignities that come with incontinence we tried to laugh off as something we were all confronting in our various ways. But I never got over the small ways Jayne's deterioration manifested itself. Like the day I realised she'd lost the use of her right arm and what this meant for a right hander; and the fact that she could no longer cut her own food or lift a cup to her lips. Or the days some small, insignificant piece of food or drink would 'go down the wrong way' and Jayne's futile efforts to cough would be enough to have me reaching to phone an ambulance. To this day, the surest form of torture for me is to watch Jayne trying to cough or struggling to breathe and not choke. It is my real and living nightmare.

Jayne has always had an excellent group of treating specialists who have supported her – and us – with compassion and honesty. Over the last 12 months, it became increasingly clear that Jayne was reaching a point of no return. She weighed barely 49kgs and was often spending up to 18 hours a day sleeping in bed. She needed help for everything and could no longer move unaided in any way.

Despite her failing health, Jayne's determination to be directly involved in the lives of her sons never wavered. She has always been fiercely independent and none more so than now.

Pompe's Disease has not only cost Jayne her health. It also destroyed her marriage and deprived her sons of their father. Pompe's Disease has ruined Jayne's career, destroyed her independence and forced her into a life of poverty and pain that has been almost impossible to witness.

But after many years of hopes being dashed as one promising treatment after another failed to fulfil its promise, we now have a life saving treatment in Myozyme. The sad thing was that by this point, Jayne had given up any hope of accessing this wonderful Enzyme Replacement Therapy as promised trials and their start dates seemed to be endlessly delayed.

It is impossible to describe the overwhelming sense of gratitude and relief we all experienced when we heard that Jayne was to be included on the Special Access Program. Jayne was numb with shock and sometimes says she still cannot quite believe it is happening.

The physical and emotional change in Jayne since she started receiving Myozyme once a fortnight has been nothing short of miraculous. Before she began the treatment she had a permanent grey pallor to her skin, was constantly exhausted and was more or less bedridden. Every day was a struggle that was becoming harder and harder to sustain. She was in enormous pain and had a very poor quality of life.

Since starting Myozyme, however, we have watched a miracle unfold before our eyes. The grey pallor has disappeared and been replaced by a healthy rosy glow to Jayne's skin. Though early days yet, the muscle on her face is slowly coming back and her breathing mask fits better. Her constant headaches have reduced in frequency and intensity and she experiences slightly less pain in her body overall.

Jayne's sense of well being is dramatically improved and though she still suffers from extensive fatigue, she now faces the future with hope and a sense of possibility she has not had since she was diagnosed. Jayne says it is as if her body is slowly awakening from a long, long sleep.

The other affecting result has been in Jayne's sons. From a situation in which they were watching their mother deteriorate before their eyes, they now have hope and confidence that their mother will be around. They still ring her several times a day from school to make sure she is alright but there is now a lightness and relief about them that is a delight to see. They now face the future with a confidence they have never had before and their wish is to have their mother back as they remember her.

Jayne has promised 19 year old Dan that she will surf with him one day and 17 year old Mac that she will dance with him at his 21st. They're promises she intends to keep!

As for the future, there are more Australians and Pompe's Disease sufferers in other countries who deserve the same chance of life that Jayne and her boys have been given. We are all determined to see this wonderful, life saving treatment made available to each and every one.

Bronwyn, Jayne's sister

Before enzyme replacement therapy (Myozyme)

First treatment with Myozyme

6 treatments later… and counting!