We have always tried to be positive

SUE AND GEOFFREY HAVE HAD TWO CHILDREN DIAGNOSED WITH NIEMANN-PICK TYPE C DISEASE. WE HAVE ALWAYS TRIED TO BE POSITIVE AND LIVE LIFE TO THE FULL, ENJOYING WHAT EACH DAY HAS TO OFFER. UNTIL FOUR YEARS AGO THIS WAS ALMOST POSSIBLE. SARAH WAS A TEACHER, HAPPILY MARRIED WITH A CHILD OF TWO AND A HALF. JONATHAN WAS IN A LONG TERM RELATIONSHIP WITH HIS GIRLFRIEND, ABOUT TO GET MARRIED AND THEN… THE STORY OF THE LAST 37 YEARS FOLLOWS.

Our daughter, Sarah

Sarah was born in June 1971. She was quickly diagnosed with liver disease and although of low birth weight and very frail, she grew into a lovely, happy girl. At 13 years old and suffering from a chronic cough, Sarah was diagnosed with Niemann-Pick type C disease (NP-C). Our son, two and a half years younger, was then tested and was also found to have this terrible disease. We were told that NP-C was a degenerative neurological condition with a fatal outcome.

> “Although both children were physically well and also doing well at school, it was awful to have such a dark cloud hanging over our heads”

Sarah's sudden death

Sarah became a primary school teacher and taught for over 13 years. Although she never presented the typical symptoms of NP-C, the knowledge that she had this illness had a profound effect on her life. She would often break down, upset that she might not live to go to university, get married, see her children grow up etc. For us it was an immense burden to watch our daughter live with these fears.

At 32 years old she married and later had a beautiful son. When she was pregnant with her second child, a little girl, she picked up a throat infection and died suddenly from septic shock.

> “Having lived for 37 years in the shadow of NP-C it seemed cruel that she should die from an infection”

Our son, Jonathan

Jonathan has had a very different experience. He was a bright child who did well at school and at sport. But when he was 16 years old he began to struggle with school work. His teachers accused him of being lazy and not bothering to work. The truth was, however, that he could not concentrate or apply his brain. He left school at 18 years old having failed his A-Levels and faced difficulties finding employment.

Jonathan eventually found work in retail, doing simple tasks. Today, he is finding these tasks increasingly challenging. One of his biggest problems is that most people don't understand why he is unable to follow or remember instructions. His condition doesn't leave any visible marks; he looks perfectly normal on the outside. As a consequence, his environment is not as patient and sympathetic as it might otherwise be.

> “Even his school friends seem unable to understand why he is doing menial jobs when he was once a bright student”

The outlook for Jonathan

Living with NP-C is not easy for Jonathan: he had to move back home, has no girlfriend, very few friends, no social life and a very unfulfilling job. He has very little money and would find it difficult to live on his own. He faces progressive deterioration in his condition. He frequently falls and injures himself.

In many aspects of his life, he now relies on our support. Understandably, he is very anxious about what is going to happen to him when we are no longer around to help him.

> “He is 38 years old and has no hope of a happy future”

NP-C is indeed a strange disease – in some respects Jonathan functions well. He has an almost obsessive interest in football and knows the names of managers and players of many teams, together with transfer values. His understanding of geography is still good yet he struggles with some of the simplest things.

NP-C and the future

In our experience, living with NP-C is distressing for all concerned – the person diagnosed with the condition, the immediate family and friends.

> “We have, however, always tried to be positive and live life to the full, enjoying what each day has to offer”

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