

## Unique Family Stories

### Jenny

Our daughter Jenny's premature arrival seven weeks early in February 1990 came as a huge shock. I had had a pretty awful pregnancy, with bleeding throughout and polyhydramnios (excess amniotic fluid) in the weeks leading up to her birth. I was absolutely enormous and was in great discomfort. Now I know that these are often signs that something is amiss with the baby but, despite many tests and scans, nothing unusual was found.

Jenny was born after a very easy delivery. In fact I hadn't even realised I was in labour until Jenny was well on her way. When Jenny was born, Tina, the midwife, just showed me Jenny's lower half so I could see I had got a girl and then Jen was whisked away by the paediatric registrar to the special care baby unit (SCBU) without me holding her or seeing her face. At first I just assumed that she needed to be in an incubator quickly but after about an hour of not being told anything, I was sure that something terrible must have happened to Jenny, maybe she had even died, and I asked Tina what was wrong. I remember her saying that she thought Jenny had a cleft lip. This was quite a relief and I said "Is that all? We'll be able to fix that." Little did I know! When I did get to see Jenny I realised that the cleft was very extensive and involved her whole palate, her nose and her upper front jaw, as you'll see from this picture. You can imagine the shock we felt on first seeing Jenny but one look at her big, dark blue eyes and my heart melted. I just wanted to hug her and make it all better.



### Jenny age 7 years



Over the next week, Jenny had to stay in SCBU while feeding was being established and I stayed in the maternity ward. The day after I went home, having to leave Jenny in SCBU, I had a telephone call from one of the nurses saying that the Consultant Paediatrician would like to meet me and my husband Trevor that evening. The consultant had been on holiday during Jenny's first week and this was his first day back in the hospital. I remember thinking what a friendly gesture this was, not imagining that he would be telling us even more bad news. How naïve I was! Trevor and I were sitting in SCBU tube feeding Jenny, me holding Jenny and gently talking to her, when the consultant came in. He asked everyone else to leave, which I thought was a bit strange. He sat down and said he had some very bad news to tell us. Jenny had a rare chromosome disorder. Up to this point we had

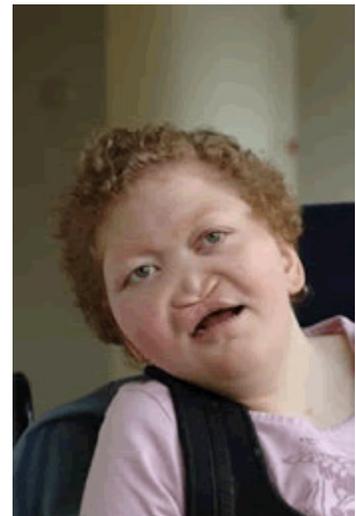
dismissed the thought that anything genetic would be wrong with Jenny - lots of children are born with a cleft lip and palate and Jenny was premature as well. We hadn't even been told that blood samples had been sent off for testing (something I am sure would not happen now). The consultant was trying his best to be gentle with us and no doubt said lots of good, consoling things but all I remember him saying were things like "Jenny will never be able to walk", "Jenny will never go to university", "Jenny will never....." In the end I asked him to just stop and leave. I couldn't take any more. As I rocked my baby, tears streaming down my face, it literally felt like I had been

smashed in the face with a sledgehammer - the physical pain was that real. It felt like my hopes and dreams for my baby were melting and draining away through my feet. How we got home that night I never know. I had driven to the hospital alone in my car and Trevor had come straight from work in his. When we got home we had to break the news to my Mum who was looking after our son Jonathan who was then just a week off his third birthday. We were stunned.

The next few days were a whirl but we decided we could either sink or swim and we made the conscious decision that the best way we could help our little girl was to get proper information and support. Well, the nurses and doctors and social workers were wonderfully supportive on an emotional and practical level but nobody could really tell us what all this meant for Jenny's future. There were some horrible, frightening articles in the medical literature listing all the possible physical and medical disorders associated with Jenny's 18p deletion, and I already understood about genes and chromosomes because I had been a microbial geneticist professionally, but nobody could tell us what Jen's life would really be like if she survived to grow up. We desperately needed to speak to someone who understood what we were going through.

### Jenny age 15 years

None of the professionals seemed to know of any relevant support groups. None of our family or friends had any idea of who we could turn to. I remember sitting on the stairs at our old house, telephone directory in hand, and just ringing as many numbers as I could think of and eventually ending up talking to someone at the Down Syndrome Association - Down Syndrome was the first chromosome disorder that sprang to mind. The chap I spoke to was very apologetic that he couldn't help but he said he had heard of a lady in Harrow who was running a small group then called "The Rare Unspecified Chromosome Disorder Support Group" and gave me her number. This was of course Edna Knight, Unique's Founder, and I thank God that I found her because we have never looked back since. Words cannot describe how wonderful it was to talk to someone who really understood what we were going through, even though our daughters had very different chromosome disorders and symptoms. What really mattered was that we felt we belonged somewhere and were no longer isolated and different from every other family around us.



From that first conversation with Edna, we could move forward and accept what life had thrown at us. Since then, we have had some really difficult times with Jenny. In our case, the consultant got it right when predicting Jenny's abilities but what he couldn't predict was the wonderfully sociable young lady she has become, despite her profound disabilities and very complex health needs. She has a great sense of humour and fun and we love her to pieces. It's because of Jenny that I have become passionate about helping families who find themselves in the same situation that we were in back in 1990 when Jenny was born and why I have ended up as CEO of Unique!

My biggest worry now is that Unique won't have enough money to keep going and to develop the services we can offer to newly-diagnosed families. It would be an absolute tragedy if all the hard work that we have put into building up the help we can give to families, and the information we hold on the database, was lost, just for the want of funding. Please share in my passion and help Unique in whatever way you can. Don't let families go back to the dark days of isolation that we experienced.