Unique Family Stories

Nell

Our daughter Nell was born with something called truncus arteriosus, a major but correctable heart defect. She was diagnosed when she was a day old. In the panic and the blind terror that ensued, I only very vaguely remember someone at the hospital mentioning something called a FISH test. I have no memory whatsoever of anyone using the words ‘gene’ or ‘genetic’, though I suppose they must have done.

Anyway: forward by four weeks. We get a phone call from Nell’s paediatrician, asking us to come in. We assume that this is a routine appointment to check her over, since she is technically in heart failure (the surgery, which took place when she was three months old, was at this point weeks away). We saunter well, kind of to his office. He checks Nell over. She is doing well. Then he sits us down.

At some point during the conversation that followed, I did what is for me a very odd thing: I stopped listening. If I could have stuck my fingers into my ears like a child, I would have. The doctor, God bless him, seemed nearly in tears. He explained that the FISH test came back and that Nell had tested positive for 22q11 deletion, aka DiGeorge Syndrome, aka VCFS. He wasn’t familiar with the condition, so he’d done what we’ve all done at some point, and gone online. He started reading to us from a huge thick pile of stuff he’d printed out. ‘There are 120 possible manifestations,’ I remember him saying. I didn’t think I’d heard properly, but one look at my white-faced boyfriend told me that I had (actually, the doctor was wrong: to date, there are 180). ‘Facial characteristics’ he said. I looked at beautiful Nell, peacefully asleep in her car seat, and wondered if it were possible for a person to go mad with grief on the spot. We’d just about computed the heart stuff. We’d forced ourselves to face the fact that open-heart surgery on a three-month-old baby is not, shall we say, risk-free. We felt scared all the time: every single minute of every single day. And now this. It was simply too much to compute.

‘Immunodeficiency problems absorbing calciumbones postural abnormalities feeding problems speech and language problems sub-mucal cleft mild to severe learning disabilities deafness... absent or malfunctioning kidneys.’ He just went on, and on, and on. ‘Seizures, strokes, bipolar disorder, scoliosis, spina bifida’ I remember staring out of the window and willing him to stop. But there was more, much more about twenty minutes’ more. Eventually, the terrible list ended. ‘She’s going to be a very special child,’ the doctor said, with a sad look on his face. ‘She is already a very special child,’ I snapped. ‘She is extremely special to us.’ ‘Of course she is,’ the doctor said. ‘But I was talking about special needs.’

Afterwards, we stood on the pavement outside the surgery and just sort of stared into space as the traffic went by. We were speechless. That night, we left Nell with her aunt and went to the pub and got drunk, and cried.

The next morning, I went online and terrified myself out of my wits. There’s so much stuff out there, and as a parent with a new diagnosis, you have no way of

knowing what's true and what isn't, what is supposition and what is fact, what is nonsense and what is helpful. One site authoritatively informed me that children with this deletion seldom survived infancy. On another site, I clicked on an innocuous-seeming link and found myself , with no warning, staring at a child's gravestone. On a bulletin board, I was told repeatedly that my daughter's feeding difficulties would be such that she would fail to thrive. And so it went on. Horrified, I never the less carried on looking. By the afternoon, I was hysterical. At this point, thank God, a girlfriend mentioned Unique. Finding this website was like coming into a cool, calm oasis after being blindfolded in the middle of a motorway. Here was sound information, advice, and the chance to meet people in the same boat. Here were knowledgeable people who could answer my questions. As it turns out, Nell's deletion is one of the more common ones, and as is true of many 'syndromes' it is extremely unusual for a person to manifest all of the possible anomalies. A year and a bit down the line, Nell is bright as a button, hilariously funny, eats like a horse, has just started standing independently and brings nothing but joy to the lives of all who meet her. She has changed my life, and changed it for the better in more ways than I could ever describe. But one of them is this: none of us knows what the future holds. Having a child like Nell, or like your son or daughter, makes every minute of every day special. The smallest things become crammed with meaning. The rubbish that seemed to matter so much before now falls by the wayside. The sun shines brighter, the rain falls harder I don't know. It's hard to put into words. All I know is, I'm glad of it, and if I could step back in time and change anything, I don't think I would.

It's not always easy, even though I know now that, if you imagine a spectrum of difficulties, we're currently at the shallow end. Many of you visiting this site won't even have a name for your child's condition. Many of you will be hundred times more frightened, bewildered and anxious than I was. My heart goes out to you. I hope it comforts you a little to know that you simply couldn't have come to a better place, and that, above all, you are not alone.

India Knight