A carrier's journey
~ Jane Devlin

Good afternoon everyone, my name is Jane. I am forty-one years old and I am a carrier of haemophilia A.

I have one brother affected by haemophilia A, one who is not, and a sister who is not a carrier.

I have a daughter who is twenty-two months old. It was the wish to have a child that originally led me to genetic counselling, and certainly that inchoate vision has been given marvellous shape in the life I now live with my daughter and her father. But this outcome followed on from the initial benefits of the counselling I received – the self-understanding, self-acceptance and peace that it instilled in me.

I only established for sure that I was a carrier of haemophilia A in 2006 after a two and a half year journey that began in early 2004. It began with yet another confession – this time to a colleague over a work dinner - that yes, I wanted children, but it was complicated for me as there was a possibility that I could have a child affected by haemophilia.

Whenever I had spoken of this to anyone previously, I had always wished afterwards that I had not. I had often experienced a sense of futility, partly to do with the fact that no-one seemed to know all that much about haemophilia and therefore either looked at me blankly, or with deep concern, or with suspicion. This last type of "look" was one that said "Oh, so of course you would never inflict that on a child... you would never have a child in that case..."

I also use the word futility because, in trying to describe what haemophilia is and what it meant through *my* personal prism for a boy or man who has it, I was talking about my brother's life, and along with this perhaps my mother's greatest achievement – my brother's very survival and the maintaining of his health – and surely these were matters far too sacred to be adequately addressed in a mere "conversation". I wanted to "talk about it" but I think, looking back now, I suppressed this need to talk, neither having found, nor expecting to find, the right forum in which to be able to do so. I think I knew that the issues arising from my family history and the likelihood that I was a carrier, dug deep into my unconscious, and that to subject them to the light of day might be to unleash a tsunami of emotion and distress.

The sense of futility also came from the feeling that no-one could understand that haemophilia did not just mean the physical realities (which, for a start, are not just the acute sort that many imagine, but, more worryingly, the chronic sort - immobilising joint pain and fears for the long-term health and quality of life of the sufferer.) In those moments of speaking to someone about it all, I knew I was trying to describe what no-one could understand unless he or she had experienced it like my brother, my mother, my father, my other siblings, or me. It was like trying to tell someone how my whole life had felt. To top it all off, in some sense I felt that this life, this story, actually belonged to my brother and my mother; it was not mine for the telling. For of course they were the ones for whom the expression of haemophilia had meant the most. It had meant a lot for my father too, but in a different way.

Now, back to that conversation with my colleague – for it was on this occasion that the response was completely surprising: my colleague had a friend who was a genetic counsellor and she would give me her details in confidence for me to pursue at will. It was such a practical suggestion in the face of all my "ifs" and "buts" about being a carrier, and certainly not one I had ever thought about. I don't think I even knew such an avenue existed for someone like me. It was a providential moment, the point of all the stumbling conversations that had gone before.

It was soon after this that I turned thirty-six, and finally began to feel ready to start exploring having a child with my partner, and therefore I was going to have to finally confront the issues around my possible, or probable, carrier status.

I contacted the friend of my colleague and she set up an initial consultation with Professor Ingrid Winship – a clinical geneticist at the General Genetics Clinic at The Royal Melbourne Hospital. I met with them both in October 2004. We met in a small room – just chairs and a table with a box of tissues on it. I already felt I was a carrier from a blood test that had been conducted some twenty years previously. I say "felt" as I remembered a figure of 92% associated with the likelihood of my being a carrier. So I knew there was some uncertainty to it, although I did not know the reason for this.

I did not need the tissues at that meeting; in fact, I felt an unlooked for sense of peace. I think this was partly because I was finally acting on something about which I had always needed to know. Until then, I had suppressed this need as I had feared any knowledge that would expose me to difficulties, for example, finding out I was definitely a carrier and what this would mean for having children.

I believe that the peace I felt at this meeting was also the result of being in an environment that was devoted to my story and to helping me with my future. I felt so lucky to have the opportunity to express myself and to be offered such specialised support and in such a professional manner.

It was just an hour or so but that hour gave me much clarity and a way forward. I was on the path of finding out about myself in terms of what impact being a carrier could have on my life; the path of action towards realising the dream of becoming a mother.

In order to review my carrier status, I was referred to Ivan Macciocca, a genetic counsellor, at The Alfred. We had our initial meeting in February 2005.

I suppose it was the day for reckoning and for removing the glad-saying from the equation. I really needed the tissues this time. I remember feeling grief for my brother and my mother and my father and the years of worry and struggle to help and treat my brother.

My brother is an amazing and utterly beautiful person, so I need to tell you that his life has also been an incredibly positive and blessed journey.

I also felt grief for the fact that for me to have a child was already so much harder, I felt, than for those without my family's genetic history. Then there was the fact that my partner was opposed to having any discussions about us having children. How was I going to convince him to go ahead with what I wanted (which was to try and have a baby) when I might have a child the care of whom might mean I could not work, and who might need frequent hospitalisation and constant treatment, and who would not be able to play football?

Anyway, Ivan was wonderful; he was an oasis of help, understanding and very practical information in the face of my sadness. It was in the formal space of the counselling relationship that the tectonic plates of my past finally shifted, releasing all the knots and pressure that had built up over my life. I cannot explain to you exactly how this shift occurred; I believe it was quite simply the counselling process, and Ivan's particular giftedness, at work.

Ivan counselled me as to all the choices I had, both in regards to finding out my status – did I even want to undergo testing to determine this - and in regards to what I might do with or without such test results.

As part of the unprecedented clarity of thought that the counselling was giving me, I wanted to know once and for all whether I was a carrier of haemophilia A. As well as the other reasons I have given for coming so late to the investigation of the issues around possibly being a carrier, another important one was not having been in relationships that were geared towards children. I think it had been convenient for me not to discuss having children and certainly I had received no encouragement otherwise from any of my partners. But then I turned thirty-six, and suddenly I discovered just how perfect a trope "biological clock" actually is; it was as if I just looked up one day and there was the clock and it was about to strike midnight.

I had always felt that, if I *were* a carrier, I would not allow this to be a barrier to having children; it would be something I would just have to deal with. But this stance was starting to be associated with guilt; I felt in some way it would be extremely selfish of me to risk having a child with haemophilia, and, therefore, to make a decision on the child's behalf that I would allow him to enter the world with an extra affliction (apart from the already numerous difficulties that being a human soul navigating this mortal coil entails.)

All of this convoluted feeling was coming out as I knew that I must find out my status.

My blood was tested. My factor VIII levels and my factor VIII antigen, coupled with my starting risk of 50%, translated as an 80% chance of being a carrier.

The main step of properly establishing the nature of the genetic change in my family was next. If a change were to be identified, my blood could then be tested for this change.

My brother agreed for his personal genetic file to be made available to Genetic Health Services Victoria, and to be tested for the specific gene change in our family. Ivan made a time to see both my brother and me at Royal Children's Hospital, to talk this through with my brother. My brother was very supportive of me and my wish to have a child, one reason being that he had always wanted to become an uncle.

The investigation of the gene change in our family would have cost about $1200 at that time, but, as it happened, there was some research already underway in Adelaide which meant that the researchers would be able to use this exercise as part of a larger project; the investigation would be free, but instead of taking two to three weeks for a result it would take up to six months.

A result finally came through and I met with Ivan in April 2006 at Royal Children's Hospital where I had a final blood test to go ahead with the comparison.

This comparison cost $250. Ivan called on 8th May 2006 with the result. The gene mutation present in my family was indeed present in me. I had fully expected it, having rightly or wrongly held the belief or fear for the previous twenty or so years that I was a carrier.

So in June of that year, my partner and I went to see Ivan, again at Royal Children's Hospital. This was important for my partner, for although I felt I knew everything and could have explained it to him, he wanted to learn about the facts in as untrammelled a way as possible, from a professional standpoint. Ivan drew diagrams about my family history and the way in which haemophilia is expressed genetically. He outlined very clearly the choices we had in regards to such options as prenatal testing.

Ivan also set up a meeting with Dr Chris Barnes, a haematologist and Director of the Haemophilia Treatment Centre at Royal Children's Hospital, and Janine Furmedge, Haemophilia Nurse Coordinator, who would give us an idea of the realities for a child born with haemophilia A today, for example: the treatments available and the prognosis, in contrast to what I had witnessed in the past.

The main thing I took away from this meeting was how amazing both my mother and my brother had been in what they had achieved in regards to my brother's general health and that of his joints, given the treatments and knowledge that had been available in my brother's childhood in the 1970s.

The outlook for me as a mother and the prime carer and the treatments available for my potential child from the start of his life were now so very much improved.

Chris also pulled no punches and did clearly state that haemophilia was still a potentially life-threatening condition.

I am sure the phrase “life-threatening” would have had much more impact had I already been a parent at that time. In any case, my partner and I decided to go ahead and try to get pregnant naturally and to accept whatever would be. This was not in the end a difficult decision: throughout the course of the counselling, I always knew I would not choose the option of assisted reproduction technology with preimplantation genetic diagnosis. To me, this would have been too much like choosing against my brother.

I had had a very important conversation with my brother, during which he had stated that he was so grateful for the chance to have been alive, and to have experienced life - a great gift - in spite of the difficulties he had faced, and even if it had turned out that his life had been only very short.

I felt then as I do now that although one would never *choose* to have haemophilia or to wish it for one’s unborn child, life *with* haemophilia may still be as fulfilling and meaningful and powerful as life without it.

I fell pregnant in March 2007 and at the twenty week ultrasound scan it was confirmed that I was carrying a girl. It was almost a shock, having done so much work to prepare for the possibility of a boy with haemophilia. Charlotte Emmanuelle was born on 10th December 2007 and is greatly loved by all, and especially by her Uncle Mike.

The process of facing the facts of my life during the genetic counselling I received was one of the most positive things that has ever happened to me. There was no pushing, nor judgement of me or my relationship, the time it had taken me or the age that I was, only the calmest and most helpful support that I could ever have wished for.

It was an experience for which I am deeply grateful. It left me feeling ready to embrace the future which was not something I could ever remember feeling previously. I believe that Charlotte was literally born out of this new hope for life. The counselling team, especially Ivan Macciocca, is to thank for this.