

The impending medical revolution in haemophilia care: one patient's view

Adam R Jones

The history of haemophilia care has been fraught with extreme successes and epic failures. The development of plasma-derived concentrates made prophylactic treatment and home care possible, but the unintended consequences were devastating for a generation and only abated with the emergence of recombinant products. Now with the arrival of longer-acting factor concentrates and the potential offered by gene therapy, further improvements in medical and social outcomes are possible. But these new treatment approaches raise challenging ethical and moral issues that society must be prepared to confront.

Key words: haemophilia, haemophilia B, Christmas disease, gene therapy, long-acting clotting factor

I was born in April 1972. Six months later I was diagnosed with severe haemophilia B, which at that time was more commonly known as Christmas disease, being so named after Stephen Christmas, the first patient in whom it was described as being different from classical haemophilia (which is now known as haemophilia A) [1]. During the ensuing years I have witnessed much of the evolution of care and treatment for haemophilia in the United Kingdom.

I can recall the days when hospital admission seemed, for me at least, to be the norm rather than the exception. Many times I was placed in traction for joint bleeds – especially knee bleeds, a practice that, as far as I am aware, has long been abandoned. As a child I remember when it was necessary to travel to hospital for treatment each time I had a bleed, and hoping that I would get there before the haemophilia unit closed so that my parents and I would not have to wait hours for the doctor on call to arrive. It made an incredible difference when I was allowed to begin home treatment and my mother was trained to prepare and administer my clotting factor.

I also remember the isolation I felt at school for being different to everyone else; this difference being most evident during physical education lessons where all the other boys in my class were learning how to play football, rugby, hockey, or cricket. In those years these were all games in which I was never allowed to participate for fear of injury.

The 1980s and 1990s ushered in the beginning of a



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purge among the haemophilia community, with lives being lost or affected by HIV/AIDS and hepatitis C [2] through no fault of their own. Not a day goes by that I don't think of my dear friends who are still struggling with, or were unable to win the battle against these dreadful diseases, which contaminated the UK supply of blood and blood products [3]. I am one of the lucky few to come through this period of devastation relatively unscathed. I don't know how, but to this day I remain HIV negative. Maybe I am CCR5 $\Delta 32$ homozygous [4]? Even so, I have contracted both hepatitis B and hepatitis C through treatment with infected clotting factor IX (FIX), but on both occasions my immune system has managed to clear these viruses without the need for medical intervention (other than a liver biopsy).

Enter the recombinants

Some years later I was elated to learn that there was a new treatment, recombinant FIX (rFIX) [5], being made available.

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The Impending Medical Revolution

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