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 plasmaderived 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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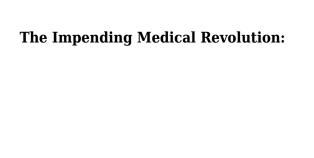
purge among the haemophilia community, with lives being lost or affected by HIV/AIDS and hepatitis C [2] through no fault of their own. Not a day goes by that I don't think of my dear friends who are still struggling with, or were unable to win the battle against these dreadful diseases, which contaminated 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 Δ32 homozygous [4]? Even so, I have contracted both hepatitis B and hepatitis C through treatment with infected clotting factor IX (FDX), 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,

The Impending Medical Revolution

SA Adler



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Table of Contents The Impending Medical Revolution

- 1. Understanding the eBook The Impending Medical Revolution
 - The Rise of Digital Reading The Impending Medical Revolution
 - Advantages of eBooks Over Traditional Books
- 2. Identifying The Impending Medical Revolution
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
- 3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an The Impending Medical Revolution
 - User-Friendly Interface
- 4. Exploring eBook Recommendations from The Impending Medical Revolution
 - Personalized Recommendations
 - The Impending Medical Revolution User Reviews and Ratings

- The Impending Medical Revolution and Bestseller Lists
- 5. Accessing The Impending Medical Revolution Free and Paid eBooks
 - The Impending Medical Revolution Public Domain eBooks
 - The Impending Medical Revolution eBook Subscription Services
 - The Impending Medical Revolution Budget-Friendly Options
- 6. Navigating The Impending Medical Revolution eBook Formats
 - o ePub, PDF, MOBI, and More
 - The Impending Medical Revolution Compatibility with Devices
 - The Impending Medical Revolution Enhanced eBook Features
- 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of The Impending Medical Revolution
 - Highlighting and Note-Taking The Impending Medical Revolution
 - Interactive Elements The Impending Medical Revolution
- 8. Staying Engaged with The Impending Medical Revolution
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers The Impending Medical Revolution
- 9. Balancing eBooks and Physical Books The Impending Medical Revolution
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection The Impending Medical Revolution
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine The Impending Medical Revolution
 - Setting Reading Goals The Impending Medical Revolution
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of The Impending Medical Revolution
 - Fact-Checking eBook Content of The Impending Medical Revolution
 - $\circ \ Distinguishing \ Credible \ Sources$

- 13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
- 14. Embracing eBook Trends
 - Integration of Multimedia Elements
 - Interactive and Gamified eBooks

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