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A five year old from Canada, born to British parents, hit the headlines in the Christmas issue of the British Medical Journal in 1952. Appropriately, his name was Stephen Christmas (whilst his brother was called Robin), and a rare blood disorder had just been named after him. Some thought it frivolous to use a Christian festival for the name of a disease; the lead author remarked that “everybody read the article because they thought it was something to do with overeating”. But this was no joke: Stephen gave his name and his voice – and ultimately lost his life – to blood disease.

Blood clotting

From the age of 20 months, it became clear that Stephen’s blood didn’t clot properly. He bled and bruised easily, often injuring himself on his toys. During a visit to England, a sample of his blood was sent to Dr Rosemary Biggs and Professor R.G.Macfarlane at the Oxford Haemophilia Centre; blood mixing experiments revealed that Stephen’s blood lacked an important protein, which soon became known as the ‘Christmas factor’, and his disorder Christmas Disease [5]: a “legitimate, unassuming and pleasantly provocative term”, said Dr Biggs.

Today, Christmas disease is known as haemophilia B [6], a rare inherited bleeding disorder. Five times more common is haemophilia A, caused by shortage of a different blood clotting protein.

"In haemophilia B, there's inadequate production of a clotting factor called Factor IX (the name now used for Christmas factor). Before modern treatment, haemophilia [7] was life-threatening. Patients were often wheelchair-bound from a young age because of bleeding into joints causing mobility problems” says Professor George Brownlee, a scientist who came up with a pioneering – and much safer – treatment for haemophilia B. The disease occurs in about one in 30,000 males; females are very rarely affected.

Blood contamination

Thirty years ago, in 1993, Stephen died - just before Christmas. His cause of death was AIDS-related, having

contracted HIV in 1985 through blood products used to treat his haemophilia. Blood transfusions had become in those days, in the words of award-winning author and journalist Andre Picard, the "Gift of Death [8]". The so-called 'tainted blood tragedy' ended thousands of lives prematurely through HIV or hepatitis infection; almost half of all Canadians living with haemophilia were infected with HIV.

Working with the Canadian Hemophilia Society, Stephen became a tireless campaigner for the cause of blood safety and sought compensation for victims. Tragically, his mother died from a stroke the day after discovering Stephen's antiviral medication and recognising his diagnosis. According to his brother Robin Christmas, in a later interview [9], Stephen felt that the 'burden of guilt' for his haemophilia diagnosis, and the shock of his HIV infection, contributed to her death.

In the UK, support schemes [10] are in place for those affected by the contaminated blood scandal, but applying for help can be 'demeaning and onerous'. The Infected Blood Inquiry [11] has been seeking answers, and compensation, since 2018 and is waiting to publish its final report. Launching the Inquiry in 2017, Theresa May described the scandal as 'an appalling tragedy which should simply have never happened'.

Link to royalty

Queen Victoria's family line was tragically affected by haemophilia B, and so the disease has since been called the Royal disease. Her son Leopold, Duke of Albany, died from blood loss after he slipped and fell; on 29th May, 1873, Prince Friedrich [12], son of Princess Alice and grandson of Queen Victoria, fell through an open window and died from a bleed to the brain.

Queen Victoria was, unknowingly, a carrier of haemophilia. She passed on the disease through her daughters, Beatrice and Alice, to the Spanish and Russian Royal families. The defect was finally discovered in 2009 by genetic studies of DNA of Victoria's granddaughter, Tsarina Alexandra, and her affected son Alexis and youngest daughter Anastasia (killed by the Bolsheviks in the Russian revolution of 1918). DNA testing of their exhumed bones showed that a single genetic defect in the Factor IX gene caused the disease.

"Haemophilia B has had an enormous impact [13] on Western history", writes haematologist Paul Monahan of the University of North Carolina.

Lost to history?

By 1958, Christmas factor was officially renamed Factor IX, with other proteins involved in blood clotting named as Factors I to VIII. Seen at the time as a solution to an otherwise chaotic situation (with one factor being known by 14 different, unrelated names), there is now a call to 'reclaim Christmas', both when naming the protein and the disease.

Consultant haematologist Dr Paul Giangrande [14] (who worked more recently at the same Oxford Haemophilia Centre where Stephen's blood disease was first correctly identified) has called for "some of the more evocative and personal names from the past" to be restored in blood clotting medicine. Since Christmas, other blood clotting proteins had also been named after patients, including Stuart-Prower Factor and Hageman Factor. Rufus Stuart had been a farmer and lay preacher who suffered from bleeding into his joints; Audrey Prower suffered from heavy periods and significant bleeding during dental surgery and when giving birth; John Hageman's blood clotting times were abnormal. All were missing blood clotting factors, and their stories aided scientific discovery.

"Just like the classification of fine wines of Bordeaux, which was devised in 1855 but which remains in widespread use to this day, we have inherited a nomenclature for coagulation factors which was established half a century ago but which is now recognized to be somewhat outdated if not inaccurate, or, at least in some cases, positively misleading", writes Dr Giangrande, reflecting on the "impersonal list of Roman numerals" in use today.

Thirty years after his death, could Christmas make a comeback in the world of medicine?

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