Hemgenix: A One-Time Treatment For Adults With Hemophilia B

National Institutes of Health
St. Jude Children's Research Hospital
University College London

Hemophilia B is a serious genetic bleeding disorder that is a result of insufficient levels of factor IX, a protein responsible for producing blood clots to stop bleeding. Symptoms include bruising, pain, swelling and prolonged bleeding. Previously, treatment involved multiple intravenous (IV) infusions – until Hemgenix.

In November of 2022, the U.S. Food and Drug Administration (FDA) approved Hemgenix, the world's first gene therapy for hemophilia B. Hemgenix is a one-time treatment for adults living with hemophilia B who have a life-threatening risk of hemorrhage or have repeated spontaneous bleeding episodes. Hemgenix emerged from pioneering work by St. Jude, led by Drs. Andrew Davidoff and John Gray, and the University College London (UCL), led by former St. Jude post doc Amit Nathwani. It was licensed to uniQure, who partnered with CSL Behring to produce the single-dose treatment to reduce abnormal bleeding by enabling continuous production of factor IX in the liver. It uses an adeno-associated viral (AAV) vector, AAV5, to deliver a Factor IX gene variant utilizing a promoter and optimized codons developed at St. Jude. The genetic instructions remain in those cells to allow stable production levels of factor IX.

Treatment is administered through an IV, and in a single dose the body receives a factor IX replacement: a gene that is expressed in the liver to produce factor IX protein, thus supporting natural blood-clotting ability.

Hemgenix is the product of more than two decades of clinical research collaboration between government and industry. The scientific contributions of the inventors, including two National Institutes of Health (NIH) institutes, represent a significant advancement for gene therapy.

The focus of the work was on Adeno-Associated Virus (AAV) vectors - a vehicle that delivers therapeutic genetic material into a cell to treat or prevent disease. Researchers aimed to find an improved and safer way to use AAV vectors to deliver the gene therapy, as well as to discover a new method of production for the vectors. While earlier AAV vectors had shown promise and advantages for gene therapy applications, the use of AAV of serotype 5 (AAV5) and a manufacturing method for the vectors, as discovered by NIH scientists, were fundamental.

Additionally, the technology transfer programs at NIH, St. Jude, and UCL were involved with licensing and commercialization. The NIH managed the initial IP filings for the case that resulted in four U.S. patents, as well as a significant number of international patents because of the global nature of hemophilia B.

After receiving Hemogenix, 94% of people participating in the clinical trial expressed factor IX to mild to normal range. Furthermore, 63% of participants in the clinical trial reported zero bleeds in the 7–18 month period following the one-time infusion. Since FDA approval and commercialization, adults living with hemophilia B are optimistic with Hemgenix.

This story was originally published in 2024.

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