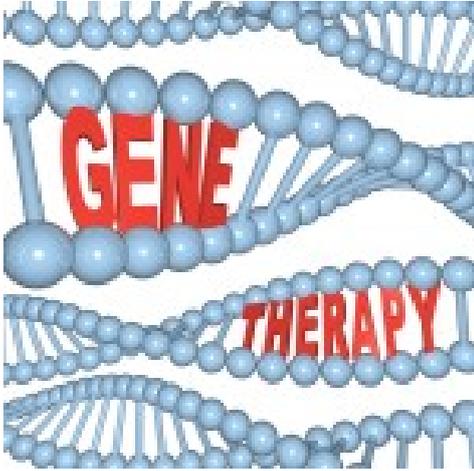


Successful Gene Therapy for Rare Hemophilia



By Lila Abassi — January 25, 2016



gene therapy via [shutterstock](#) [1]

Hemophilia was once popularly referred to as the royal disease. It was passed through the blood line of Queen Victoria to her progeny, which affected several royal families in Spain, Germany and Russia.

Hemophilia is an inherited bleeding disorder that can be either X-linked, meaning passed along on the X chromosome of female carriers primarily affecting male offspring, since males inherit only one copy of the X chromosome containing the defective gene. Or they can be autosomal recessive, where two bad copies need to be inherited for the disease to manifest. Sufferers of this disorder run the potential risk of life-threatening bleeding, as their blood cannot clot properly.

A study published in the journal, *Blood* [2] reveals how researchers were able to use a single gene therapy injection in dogs to successfully correct a rare type of hemophilia. Factor VII deficiency is an autosomal recessive bleeding disorder that affects about one in 500,000 people.

There are two types of [Factor VII deficiency](#) [3], Type I and II, with the former being the most common. People with this disorder can have bleeding into the joints, the gastrointestinal tract and the central nervous system, nose bleeds, bleeding into the muscles, heavy periods and significant post-operative bleeding.

Scientists were able to utilize dogs with the same type of hemophilia to correct the factor VII deficiency. The research team used the adenovirus (virus that causes the common cold) as a carrier to inject the dogs with the corrected version of the Factor VII gene. Within three months of receiving the injection, the dogs had a level of Factor VII in their blood that would be considered therapeutic in humans. Additionally, the injection had a very safe side-effect profile, as evidenced by the dogs' normal kidney and liver enzymes no toxicity was observed, nor were there any issues with excessive clotting.

Our finding has great clinical relevance for patients with factor VII deficiency, stated lead author, [Paris Margaritas](#) [4], DPhil, Research Assistant Professor of Pediatrics at the Raymond G. Perelman Center for Cellular and Molecular Therapeutics (CCMT) at the Children's Hospital of Philadelphia. These dogs have a type of mutation found in the majority of patients with this disorder, so this approach could lead to a sustained gene therapy in people, she added.

[Genes](#) [5] are composed of DNA that carries information needed to make proteins the building blocks of our bodies. Aberrations in the DNA sequence or code of a gene are called mutations, which often are harmless but sometimes can lead to serious disease.

The principle behind [gene therapy](#) [6] is to replace a non-functioning, or a dysfunctional gene, with one that works correctly. Viruses are often used as vectors think of them as molecular delivery trucks to deliver the correct copy of a gene by infecting and incorporating their genetic material into the host cell (but the viruses themselves have been genetically engineered to not cause disease).

The vector is introduced directly into tissues (such as the liver or lungs) and each subsequent time the cell replicates it will contain the corrected genetic material. Gene therapy holds much promise. Hopefully, the success of the hemophilia study will be similarly observed once human trials begin and we will be that much closer to curing many more diseases using this method.

COPYRIGHT © 1978-2016 BY THE AMERICAN COUNCIL ON SCIENCE AND HEALTH

Source URL: <https://www.acsh.org/news/2016/01/25/successful-gene-therapy-for-hemophilia>

Links

[1] <http://www.shutterstock.com>

[2] <http://www.bloodjournal.org.newproxy.downstate.edu/content/bloodjournal/early/2015/12/23/blood-2015-09-671420.full.pdf>

[3] http://www.uptodate.com.newproxy.downstate.edu/contents/rare-inherited-coagulation-disorders?source=search_result&search=factor+vii+deficiency&selectedTitle=1~18

[4] http://www.mdlinx.com/oncology/article/313?utm_source=in-house-msg&utm_medium=message&utm_campaign=long-jan16-oncology

[5] <http://www.ama-assn.org/ama/pub/physician-resources/medical-science/genetics-molecular-medicine/current-topics/gene-therapy.page?>

[6] <http://ghr.nlm.nih.gov/handbook/therapy/procedures>