

WHAT ARE THE GOALS OF GENE THERAPY RESEARCH?

VOLUME 5.0

We typically take medicine to help us feel better, whether physically or mentally. In the case of gene therapy, clinical trials are underway to determine if, for some people, gene therapy could reduce or eliminate the need for ongoing treatment and the burdens of chronic disease. It's important to remember that the risks and long-term effects of gene therapy, including duration of effect, are also being studied and have not yet been determined.

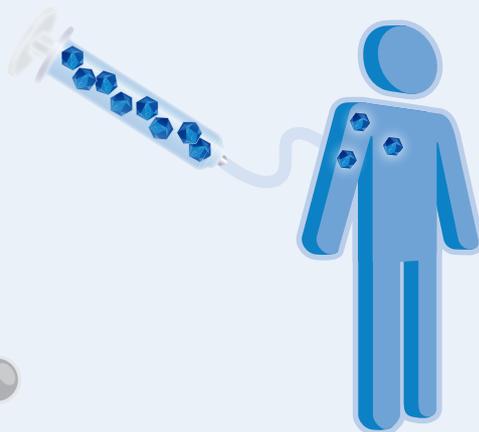
ADDRESSING GENETIC MUTATIONS

Gene therapy aims to address specific **mutations** in an individual's genetic instructions that have interfered with the body's ability to produce the **proteins** it needs. These mutations are addressed by:



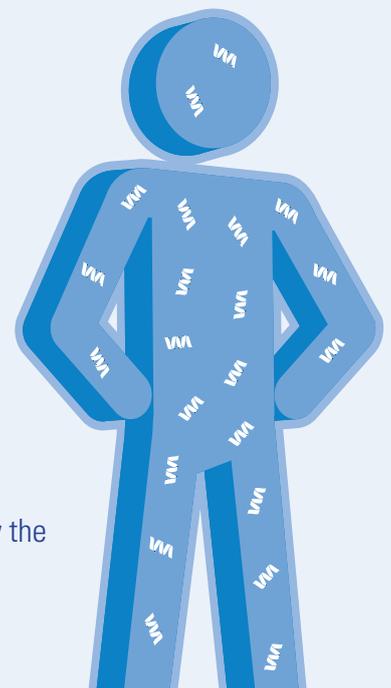
1

Introducing functional genes into the body to try to target the mutations responsible for genetic conditions



2

Producing the proteins needed for proper function. For example, in **haemophilia A** or **B**, the goal is to allow the body to produce **factor VIII** or **factor IX**, respectively



POTENTIALLY REDUCING THE NEED FOR TREATMENT

Research is ongoing to determine if gene therapy can help the body produce the proteins it needs. This may lead to less reliance on currently available medication.



POTENTIALLY ELIMINATING OR REDUCING SYMPTOMS

Clinical trials are evaluating whether gene therapy could eliminate or reduce the need for routine disease management. This in turn could possibly lessen the physical, mental, and emotional burden of a disease. Although gene therapy may not be able to address preexisting damage, it may be able to mitigate progression of any existing damage.

All of these goals (as well as the risks) of gene therapy are currently being evaluated in clinical trials.

GLOSSARY

Mutation—A change in the structure of a gene; this variant form of the gene may be passed along to future generations. It is caused by the alteration of single base units in DNA, or the deletion, insertion, or rearrangement of larger sections of genes or chromosomes.

Protein—A group of amino acids joined together that perform various biological functions. Examples include enzymes, hormones, and antibodies.

Haemophilia—A bleeding disorder that slows the blood clotting process. People with haemophilia can experience both spontaneous bleeding and prolonged bleeding following an injury. Serious complications can result from bleeding into the joints, muscles, brain, or other internal organs. The main types of this condition

are haemophilia A (factor VIII deficiency) and haemophilia B (factor IX deficiency).

Factor VII—The factor VIII gene provides instructions for making a protein called coagulation factor VIII. Coagulation factors are essential for the formation of stable blood clots. Factor VIII is absent or inactive in people with haemophilia A.

Factor IX—The factor IX gene provides instructions for making a protein called coagulation factor IX. Coagulation factors are essential for the formation of blood clots. Factor IX is absent or inactive in people with haemophilia B.

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