ASHG
American Society of Human Genetics

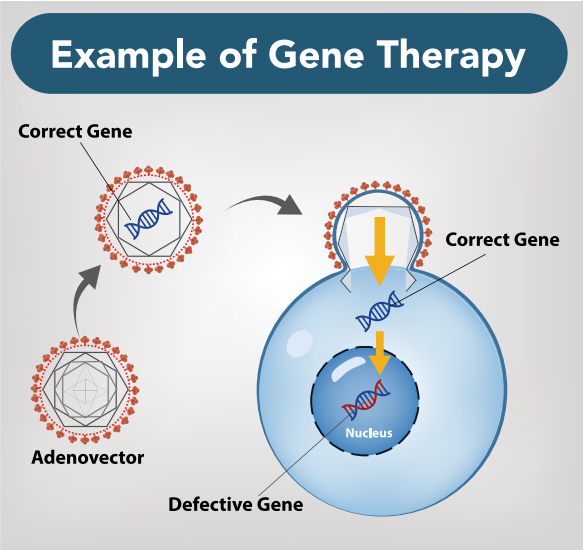
Success Stories
in Human Genetics and Genomics Research

GENE THERAPY:
FROM FUNDAMENTAL RESEARCH TO CLINICAL SUCCESS

Scientists first proposed treating disease by changing the DNA in a person's cells nearly 50 years ago.[1,2] Today, the promise of gene therapy is beginning to be realized—and it would not be possible without fundamental research supported by the National Institutes of Health (NIH).

Genes as Medicine

Gene therapy is a way to supplement or modify a person's genes to treat, cure, or prevent disease.



If you think of the cells in our bodies as machines, gene therapy allows scientists and doctors to act as mechanics and open the machines to investigate, understand, and potentially repair the internal pieces. Repairs might replace or inactivate a disease-causing gene, or add genes to help treat disease.[3,4]

Of course, this is much more difficult in an actual human cell. For any gene therapy, researchers must determine the underlying genetic cause, what corrective steps are needed to help the patient, and how to deliver it to the right cells. This requires years, sometimes decades, of fundamental research on the patient's disease, the target cells and tissues, and the gene delivery system.

Supported by NIH, researchers have spent decades studying the fundamental science underlying gene therapy approaches and how to apply them safely. Over time, scientists have developed new ways to deliver genes and a better understanding of target cells. These advances have ultimately led to the successful clinical translation of gene therapies.[5]

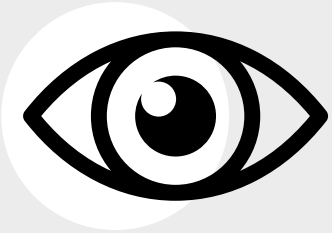
In the United States, the Food and Drug Administration (FDA) approved the first gene therapy products in 2017. These include CAR-T cells to treat blood cancers and a gene replacement therapy to treat congenital blindness.[6]

Unexpected Clinical Applications from Discovery Research

Investments in fundamental research can lead to surprising clinical benefits, alongside more targeted research efforts. For example, the genome editing tool known as CRISPR is making gene therapies more efficient, more durable, and more precise. Yet this transformative new tool was originally discovered by researchers studying how bacterial immune systems defend against viruses that infect bacteria. Now, CRISPR technology is in clinical trials to cure diseases like sickle cell disease and severe, familial high cholesterol with a single treatment.[7]

NIH-funded Research is Driving Discoveries

Leber congenital amaurosis (LCA) is a group of diseases caused by inherited gene mutations that result in significant vision loss or blindness in infancy.[10] A transformative gene therapy can improve vision by delivering a normal copy of the *RPE65* gene to cells in the retina.[11] Click here or follow this link: bit.ly/45u5yrE to learn more.



How Can Congress Support Research?

Currently, there are nearly 20 gene therapy medicines approved by the FDA and over a thousand registered clinical trials involving gene therapy.[8,9] With robust and predictable NIH funding, these numbers will continue to grow.

Decades of fundamental research were essential to the development of current gene therapies, and more research is needed to develop new and better ones for the future. While a limited number of gene therapies are in use, our knowledge is far from complete. More research remains to be done in order to improve current gene therapies and develop new ones.

Congress can support this research by increasing the funding to the NIH, which supports broad biomedical research programs that continue to be essential for gene therapy research. With sustained funding, gene therapy may one day treat a wider range of inherited diseases, cancers, and other health conditions.

Additional Resources

Gene therapy comes of age: https://www.science.org/doi/10.1126/science.aan4672

How gene therapy is emerging from its 'dark age': *https://www.nature.com/articles/d41586-022-04210-5*

What is Gene Therapy? *https://www.fda.gov/vaccines-blood-biologics/cellular-gene-therapy-products/what-gene-therapy*

How Gene Therapy Can Cure or Treat Diseases *https://www.fda.gov/consumers/consumer-updates/how-gene-therapy-can-cure-or-treat-diseases*

References: *ashg.org/advocacy/fact-sheets/*