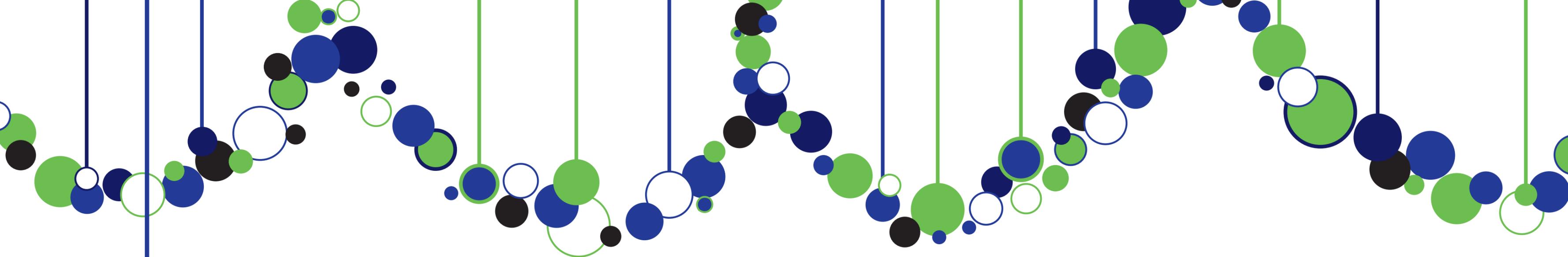


# The science of gene therapy



Spark Therapeutics, a leader in the field of gene therapy research.



## Starting from the beginning

### What is a gene?

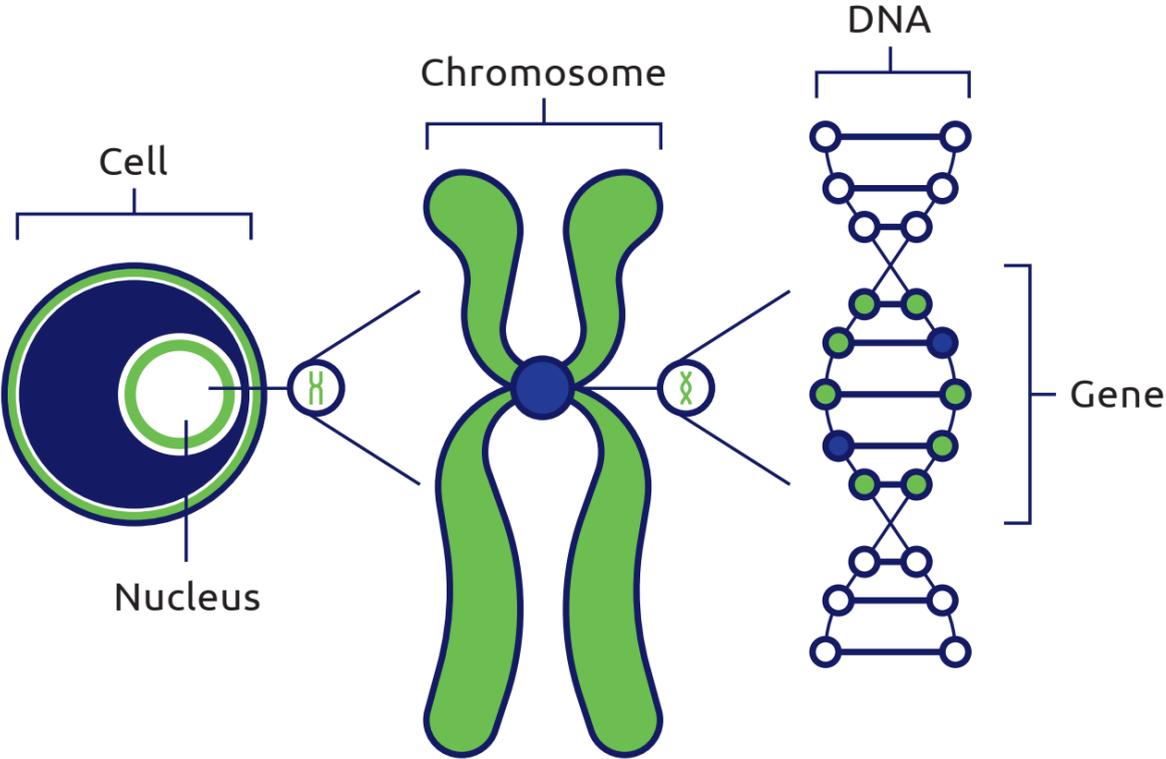
The human body is composed of trillions of cells.

Cells are the basic building blocks of all living things. The command center of each cell is called the nucleus, and it contains chromosomes. Chromosomes are made up of DNA—the body's hereditary material.

# Genes are segments of DNA

Typically, every person has 2 copies of each gene—one from each parent.

A person has about 20,000 genes, and most are the same in all people. However, each person has a small number of genes that are slightly different. These slight differences in genes are why people have different features.



*Genes contain instructions for making proteins, which are molecules that build, regulate, and maintain the body.*

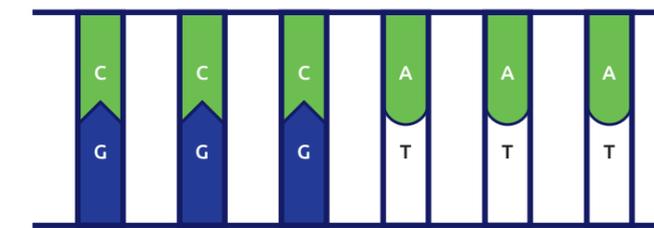
## The role genes play

### What is a genetic disease?

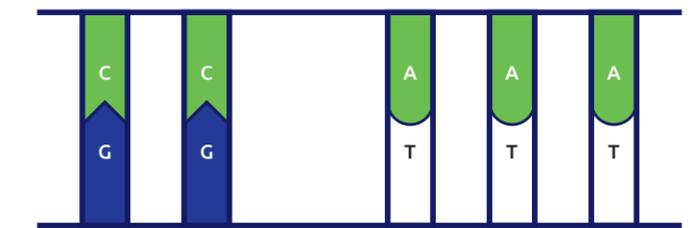
Sometimes, there is a change in a gene's DNA sequence, such as a substitution, deletion, or duplication. This is called a mutation and can cause a necessary protein to not work properly or to be missing. A mutation can be passed from parent to child or can be acquired during a person's life.

Some changes in genes are harmless, but others can affect our health. **Gene mutations can result in genetic diseases.**

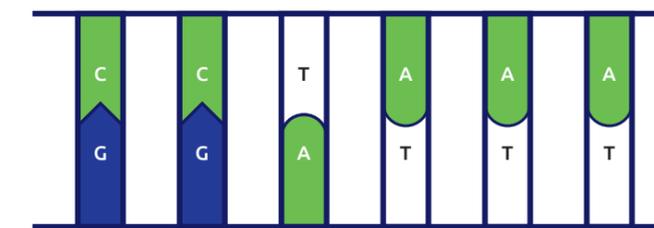
*Gene therapy research has the potential to find ways to treat diseases that were previously untreatable.*



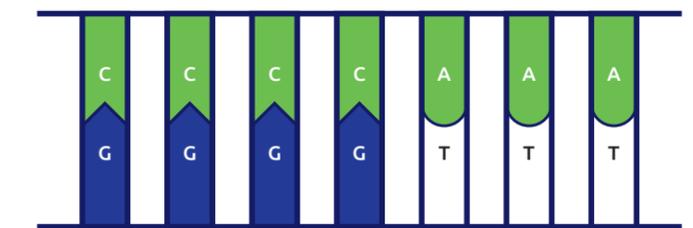
Original DNA sequence



Deletion



Substitution



Duplication



## How gene therapy fits in

### What is gene therapy?

Simply put, gene therapy is an investigational approach to treat or prevent genetic disease.

Gene therapy research is not new. In fact, scientists have been investigating and evolving it for more than 50 years.

Gene therapy research has the potential to find ways to treat many diseases; some of these diseases are partly or fully caused by genetic mutations, such as:



**Cardiovascular disease**



**Neurodegenerative disorders**  
(such as Parkinson's disease)



**Vision disorders**



**Diabetes mellitus**



**Blood clotting disorders**  
(such as hemophilia)

### Gene therapy versus cell therapy

People may confuse gene therapy with cell therapy. Both are meant to help treat disease, but they are not the same.

- **Cell therapy** transplants whole cells into a person
- **Gene therapy** manipulates cells by bringing genetic material into cells that are not working correctly or could work better

## Gene therapy in action

### What is it meant to do?

The goal of gene therapy research is to determine whether a new gene can be used to replace or inactivate a mutated gene to treat a disease or help the body fight a disease.

For a gene to be delivered into a cell, a transporter is typically used.

**A transporter is known as a vector.**

## The role of a vector

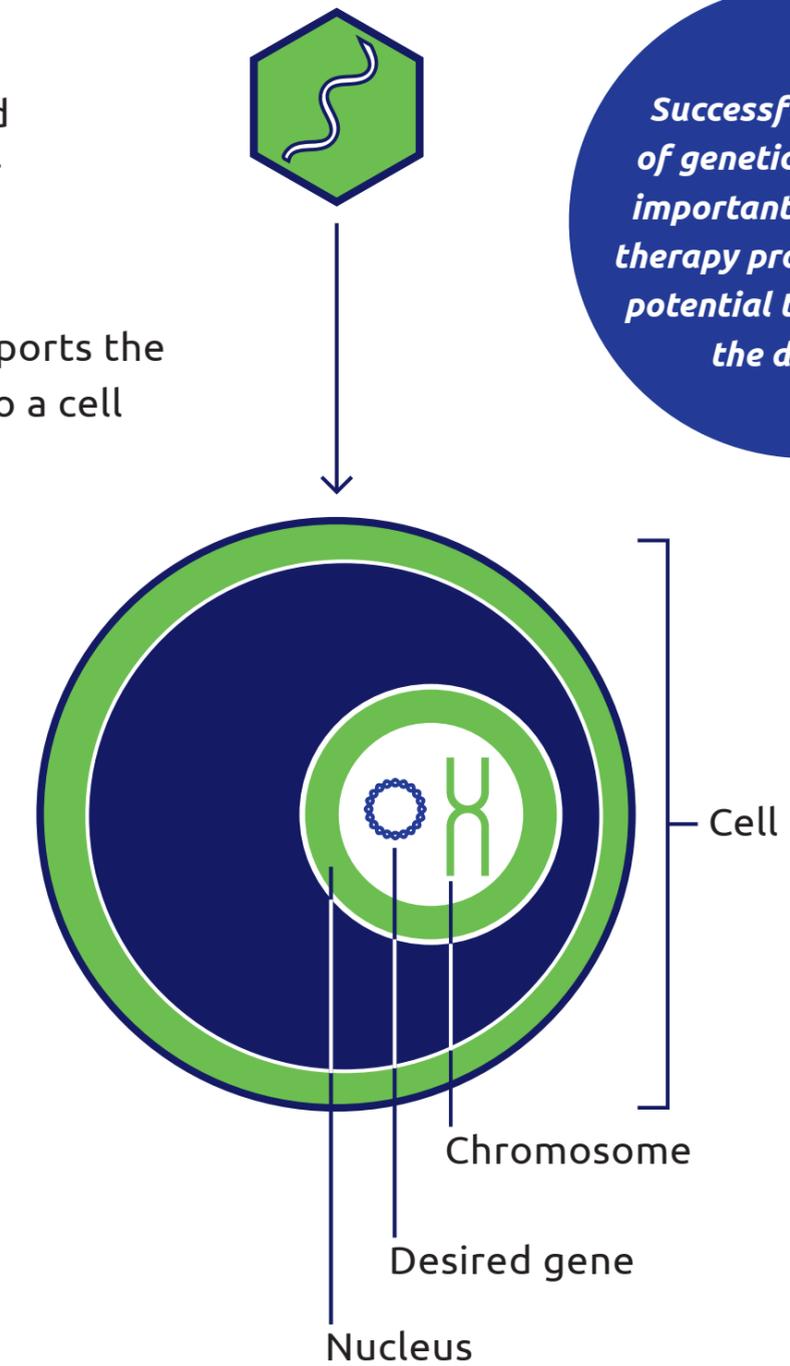
- A vector is made from an altered virus
- Before the virus is used as a vector, its viral genes are removed
- Once the virus is modified, it is intended to transport the desired gene to a cell without causing disease
- Once inside, the desired gene should restore the function of the protein

Vectors can be given intravenously (by IV), which means they are administered into a vein, or injected into a specific tissue in the body. Other procedures, such as surgery, can also be used to deliver vectors into specific areas of the body.

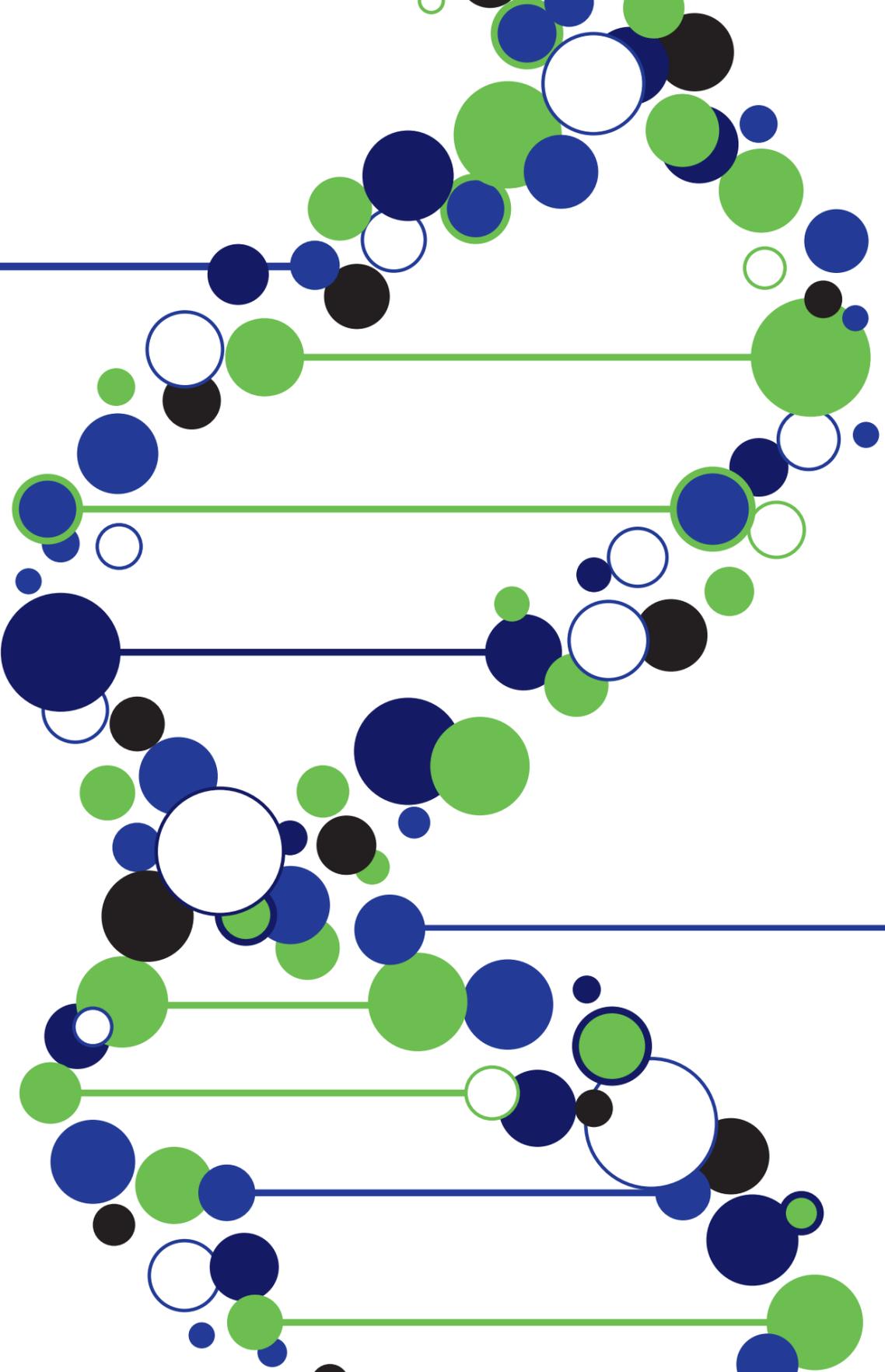
The desired gene is packaged inside the vector

The vector transports the desired gene into a cell

The desired gene works within the cell



# Bringing hope to millions



The potential of gene therapy research has given hope to millions of people impacted by genetic diseases.

At Spark Therapeutics, a fully integrated company, we work with rare disease advocacy groups from around the world to help connect patients and families to education, research, support services, and each other.

We strive to challenge the inevitability of genetic disease by seeking to discover, develop, and deliver gene therapies that address inherited retinal diseases (IRDs), neurodegenerative diseases, as well as diseases that can be addressed by targeting the liver.



To learn more about  
our research in gene  
therapy, visit us at  
[Sparktx.com](https://www.sparktx.com).

