

Understanding New Discoveries in Genomic Medicine

While the exact cause for many genetic diseases remains unknown, for some patients, the problem can be traced to errors in DNA, or changes in a person's genes that can result in rare and severe disease. Emerging research in genomic medicine is working to address the underlying mistakes in DNA that lead to some genetic diseases.

The following answers five frequently asked questions about the emerging field of genomic medicine including gene therapy, genome editing and cell therapy, and what these technologies could mean for the rare disease community.



What is Gene Therapy?

Gene therapy is the treatment of disease by delivering a new gene into a patient's cells to replace an incorrect or damaged gene. Most often, gene therapy works by introducing a corrected copy of a defective gene into the patient's cells, without removing or modifying the defective gene. The goal of gene therapy is to treat or cure a genetic disease by adding back a normal copy of the gene that was responsible for the disease.

Although the concept sounds futuristic, gene therapy has been studied by researchers for more than 40 years. Along the way, much attention has been focused on ensuring this complex technology is implemented safely and effectively.



What is Genome Editing?

Genome editing (also called gene editing) makes permanent changes to the genetic code of a cell, by correcting, disabling, removing, or modifying the DNA.

Genome editing works by using enzymes called "engineered nucleases" to make precise cuts at specific DNA sequences. With these engineered nucleases the DNA of a cell can be changed permanently.

There are several types of engineered nucleases used in genome editing. These include:

- Zinc Finger Nucleases (ZFN)
- Transcription Activator-like Effector Nucleases (TALEN)
- Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR)

Each genome editing tool has slight differences, but all engineered nucleases are designed to do the same thing: act like molecular scissors to cut the DNA at a precise spot in the genome to remove, add, or replace the DNA. After cutting the DNA, the cell will naturally repair the break and researchers use these natural repair processes to introduce specific changes at the site where the DNA is cut.





What is Cell Therapy?

Cell therapy is the infusion or transplantation of human cells into a patient for the treatment of a disease. The origin of the cells can be from the patient or from another healthy donor. This technology uses the patient's own cells or cells from a healthy donor as a therapy to treat the patient for diseases caused by mistakes in DNA, infections or cancer.

Whole blood transfusions, packed red blood cell transfusions, platelet transfusions, bone marrow transplants, and CAR T-cells are all forms of cell therapy.



Can Genome Editing and Cell Therapy Be Combined?

Genome editing and cell therapy both have the potential to ease the underlying cause of genetic diseases and can be combined for certain treatments. Cells are collected from the patient or another healthy donor, modified in a laboratory using genome editing and re-administered to the patient.

Numerous approaches are currently under investigation, with a promising therapy involving a type of cell therapy called CAR T-cell therapy. With this treatment, T-cells, a type of human immune cell, are collected from blood, modified with engineered nucleases in the laboratory to make them better at attacking cancer cells and returned to the patient.

What Does This Mean for the Rare Disease Community?

Genomic medicine is making a positive impact in the fields of oncology, rare, and infectious disease. While early research and emerging clinical trials are encouraging and offer hope to patients and their families, many more studies are needed to ensure these therapies are safe and effective for adults and, someday, children.

Hundreds of clinical trials are being conducted to test gene therapy, genome editing and cell therapy as potential treatments for some genetic conditions, certain cancers and HIV/AIDS. This is an exciting new frontier in medicine and you can visit ClinicalTrials.gov to search for clinical trials that are accepting patients. Your physician or a genetics professional can help you understand if a clinical trial may be right for you.

