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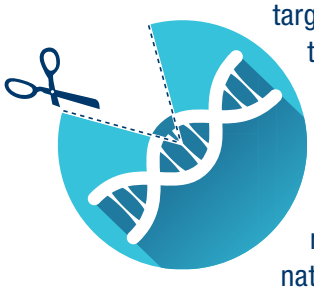
HUMAN GENOME EDITING

The genome is comprised of DNA and functions as the "instruction book" of a cell. Genes are specific strands of DNA, which provide the cell instructions for making different proteins. Humans have approximately **20,000 genes**, and there are more than **6,000 genetically based diseases**. As our understanding of genetics advances, this number will continue to grow.

How the Genome Editing Process Works

Genome editing is a process by which a strand of DNA is modified to produce a desired outcome

Genome editing is based on a naturally occurring system that directs a molecular scissor, called a nuclease, to a target region of DNA. The DNA is targeted by a recognition signal that is specific to a fragment of DNA. Once the nuclease has been directed to the appropriate region of DNA, it cuts the DNA. The DNA is then repaired by a process found naturally in each of our cells. This repair machinery is used to fix any break that may occur within the DNA. Once the DNA is cut, the cell can be directed to repair that region of DNA in three different ways:



1. Insertion of a DNA sequence, when a template DNA is provided to the cell in parallel with the targeted cutting.
2. Deletion of a DNA sequence, when two regions of DNA are cut.
3. Change of a DNA sequence, when a corrective template DNA is provided.

GENOME EDITING IS DIFFERENT THAN GENE THERAPY

Genomic medicine is a broad term that encompasses Gene Therapy and Genome Editing

Gene Therapy

Gene Therapy introduces one or more new copies of a gene into the patient's genome to restore cell function despite the continued presence of the mutated gene.

Genome Editing

Genome Editing corrects or removes a defect in a gene. Once changed, the correction will persist throughout the lifespan of the cells.

SOMATIC CELLS

Somatic cells are cells whose genetic material cannot be passed on to future generations. The vast majority of cells in the human body are somatic cells.

GERMLINE CELLS

Germline cells (or germ cells) are cells whose genetic material may be passed on to future generations; these include sperm, egg cells, or fertilized embryos.

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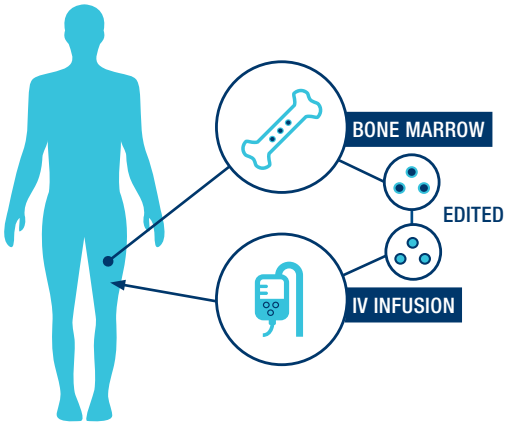
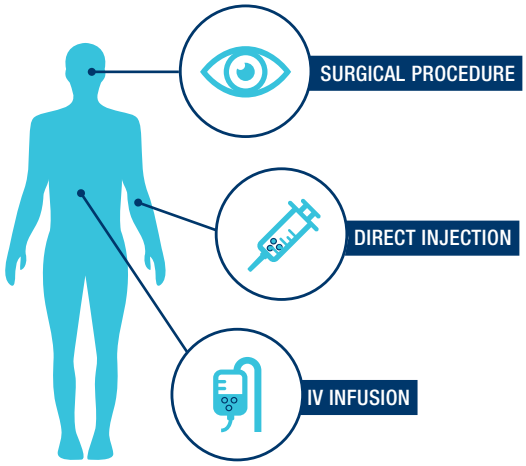
HOW SOMATIC CELL GENOME EDITING WORKS IN PATIENTS

Somatic cell genome editing can happen outside the body (**ex vivo**) or inside the body (**in vivo**). Each method has benefits and limitations, and preference of method depends on the disease being treated.



EX VIVO GENOME EDITING

In Ex Vivo Genome Editing, the target cells — for example, blood cells — are first removed from the patient. The cells are then treated in a laboratory to edit the target gene, and return back to the patient.



IN VIVO GENOME EDITING

In Vivo Genome Editing occurs inside the human body and is directly delivered to the target site — for example, a diseased liver — where the genome editing therapy would find and enter the target cells.

GENOME EDITING TECHNOLOGIES

Broadly speaking, there are three major genome editing technologies in use today to delete, insert, or repair DNA. Although each works slightly differently, each technology relies on nucleases — proteins that "cut" DNA — and they can all bind to and edit targeted genes as directed. These include:

- Zinc finger nucleases (**ZFNs**);
- Transcription activator-like effector nucleases (**TALENs**); and
- Clustered Regularly Interspaced Short Palindromic Repeats (**CRISPR**) nucleases.
- **Meganucleases** represent a fourth category, but this category is not as widely used for potential clinical applications.

Research is underway on clinical applications of genome editing technologies to treat genetic disorders like **sickle cell disease**, **cystic fibrosis**, **congenital blindness**, **hemophilia**, **amyloidosis**, and **lysosomal storage disorders**.

Learn more

For more detailed information on the U.S. Regulatory framework currently in place please visit BIO's FAQs on Genome Editing, and check out BIO's Position Statement on Human Genome Editing.