ASHG
American Society of Human Genetics

GENE EDITING:
BRINGING TRANSFORMATIVE TREATMENTS TO LIFE

WHAT IS GENE EDITING?

GENE EDITING MAKES CHANGES TO DNA TO STUDY OR TREAT DISEASE

Gene editing is a technology that changes an organism's DNA sequence. Gene editing can be used to treat certain types of genetic disorders and other conditions by precisely locating and changing the section of DNA that causes the disorder. It is also used in research to study how human diseases develop. While there are several types of gene editing technology, here, we will focus on the most widely used technology—CRISPR/Cas.

HOW IS GENE EDITING USED IN RESEARCH?

Gene editing is used in research to study the causes of human diseases. Researchers often study the effects of gene editing in animal models and in animal or human cell lines.

ANIMAL MODELS

Animal models such as mice and zebrafish share many of the same genes with humans. Gene editing can be used to inactivate a specific gene in an animal model to understand how the absence of that gene may cause disease in humans. This research helps us understand how our DNA contributes to conditions such as birth defects, heart disease, diabetes, cancer, neurological disorders, and mental health disorders.

CELL LINES

Alternatively, animal and human cell lines provide a simplified model that allows scientists to study biological processes in specific organs or tissues under controlled and standardized conditions. Gene editing can make precise changes in specific genes of the cell line. This research helps us understand how genes function and affect disease development, accelerating the discovery of new therapies.

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HOW DOES CRISPR/CAS TREAT GENETIC DISORDERS?

1 FIND DNA SECTION
Guide RNA locates faulty gene

2 CUT DNA
CRISPR scissors

3 REPAIR OR REPLACE DNA
CRISPR uses cell's repair process to fix it

CRISPR/Cas is short for clustered regularly interspaced short palindromic repeats and CRISPR-associated systems. CRISPR/Cas is a type of gene editing that uses helper molecules, called guide RNAs, to precisely find the exact section of DNA causing the disease and cut the DNA at that location. CRISPR/Cas then tells DNA's natural repair process to remove, correct, or add genetic material. CRISPR/Cas has successfully treated several genetic disorders, unlocking new possibilities and transforming patient and family lives.

The First FDA-Approved Gene Therapy

SICKLE CELL DISEASE

CRISPR/Cas is the first gene therapy approved by the U.S. Food and Drug Administration (FDA) to treat sickle cell disease (SCD), an inherited blood disorder. SCD is caused by a genetic change that makes red blood cells become sickle-shaped, which decreases the flow of oxygen to the body. CRISPR/Cas therapy treats SCD by collecting a person's stem cells (cells that can become any other type of cell in the body), editing them in the lab to replace or correct the faulty gene, then transplanting them back into the person. This increases the production of healthy red blood cells and improves oxygen delivery to the body. This marked a breakthrough for SCD and opened the door to effective treatment options for many more incurable diseases.

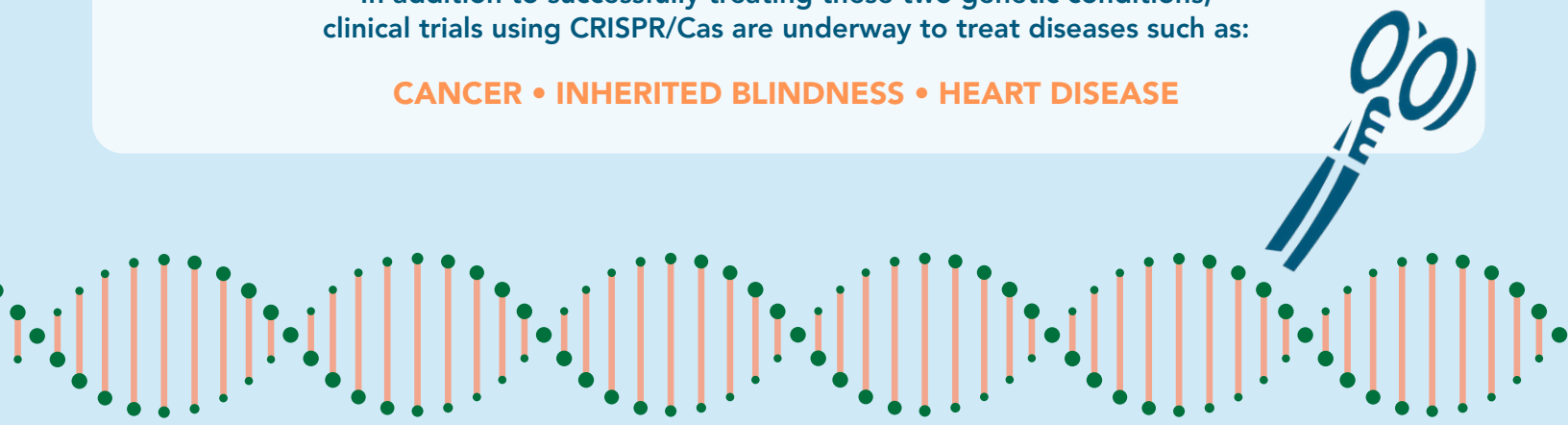
Personalized Treatment for Rare Disorders

CPS1 DEFICIENCY

CRISPR/Cas was used to treat a child with a rare genetic disorder called severe carbamoyl phosphate synthetase 1 (CPS1) deficiency, a life-threatening disease. CPS1 deficiency is caused by a genetic change that prevents the body from breaking down proteins in the liver, leading to a toxic buildup of ammonia in the body and severe brain damage. CRISPR/Cas technology was customized to correct the child's specific genetic change and, within six months of being diagnosed, the child received treatment. The therapy successfully decreased the buildup of ammonia in the body, establishing the foundation for using personalized therapies to treat other genetic disorders.

In addition to successfully treating these two genetic conditions, clinical trials using CRISPR/Cas are underway to treat diseases such as:

CANCER • INHERITED BLINDNESS • HEART DISEASE



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IS GENE EDITING SAFE?

As with any new biotechnology, there are some safety concerns. For example, unintended errors could occur during gene editing, and the potential long-term effects in humans are still being studied. However, clear guardrails are in place that evaluate the safety of gene therapies before they are used in humans.

CURRENT GUARDRAILS:

- ☑ The FDA approves treatments.
- ☑ The National Institutes of Health (NIH) has policies for all gene editing and gene therapy trials it funds.
- ☑ Human clinical trials involving gene therapy must be reviewed by an Institutional Review Board (IRB) and an Institutional Biosafety Committee (IBC).

IS GENE EDITING ETHICAL?

SOMATIC CELL EDITING: ACCEPTABLE

Gene editing of somatic cells (cells that are not passed on from parent to child) is considered an ethical form of treatment by most researchers and ethicists.

GERMLINE/EMBRYO EDITING: BANNED IN MANY COUNTRIES

There are significant concerns of potential misuse when it comes to gene editing of germline cells (egg and sperm cells) and embryos, such as enhancing certain traits that can be passed on to the next generation, also known as eugenics. Because of these concerns, clinical use of germline and embryo gene editing is illegal in the United States, the United Kingdom, the European Union, and many other countries.

COST/ACCESS: EQUITY CONCERNS

Another ethical consideration of gene therapy is cost. Currently, gene editing therapies are time-consuming and expensive. Some ethicists and others in the scientific community are concerned that some people won't be able to get these treatments because they cannot afford them.

WHAT IS THE FUTURE OF GENE EDITING?

Gene editing technology has transformed how we study and treat human disease. New gene editing technologies are continuously emerging, and international frameworks evolve with them to guide ethics and prevent misuse. In the future, FDA-approved CRISPR/Cas drugs and other gene therapy drugs will likely become routine for some rare diseases. As technology becomes faster, cheaper, and more accurate, gene editing has the potential to solve the most difficult problems in medicine, saving more lives and reaching thousands of people across the world.

LOOKING AHEAD

Human genetics and genomics research has achieved extraordinary progress, laying the foundation for innovations that improve health and well-being worldwide. Looking ahead, the next generation of discoveries – driven by emerging scientists and transformative technologies – holds even greater promise. Through continued investment, we can ensure these advances benefit all people, reflecting our broad community and global impact: One Humanity, Many Genomes™.

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