American Journal of Biomedicine and Pharmacy
https://biojournals.us/index.php/AJBP
ISSN: 2997-7177

The Possibilities of CRISPR–Cas9 Technology in the Treatment of Hereditary Diseases

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Received: 2025 06, Sep
Accepted: 2025 06, Oct
Published: 2025 24, Oct

Abstract

In genetic engineering, the revolutionary CRISPR-Cas system has emerged as a crucial tool for precise genome editing. At the same time, the emergence and rapid development of deep learning methods have provided new impetus for scientific research in genomic data analysis. Since advancements in both fields are occurring simultaneously, it is necessary to continuously monitor these rapidly evolving research directions. Significant progress has been achieved in utilizing deep learning to predict the activity of guide RNA (gRNA) within CRISPR-Cas systems. The activity of gRNA is a key factor determining the accuracy and efficiency of genome editing. By analyzing recent studies, it becomes evident that there have been remarkable achievements and new directions in integrating CRISPR-Cas systems with deep learning. This integration contributes to an important interdisciplinary field that bridges artificial intelligence and genetic engineering.

Keywords: CRISPR-Cas system, deep learning, guide RNA, genome editing, on-target activity, off-target activity, artificial intelligence, molecular scissors, DNA, mutation, genetic disorders, Cas9 protein.

Introduction

Genetic diseases are serious health problems that arise as a result of mutations in the human genome. Traditional drug therapy or physiotherapeutic methods cannot eliminate the root cause of these diseases.

The CRISPR–Cas9 technology was first introduced to the scientific community in 2012 by E. Charpentier and J. Doudna. Since then, research in the field of gene editing has expanded widely in many scientific centers around the world. To date, various organisms, including human cells, have been successfully genetically modified using CRISPR (Jinek et al., 2012).

Clinical studies have shown that this technology has helped restore vision in certain forms of hereditary blindness, such as Leber congenital amaurosis (Maeder et al., 2019). Moreover, scientific publications have reported positive outcomes from clinical trials targeting sickle cell anemia (Frangoul et al., 2021).

The CRISPR system consists of two main components:

CRISPR locus – a region in the bacterial genome composed of short, repetitive sequences separated by spacer sequences derived from viruses or plasmids that have previously invaded the bacterium (Mojica et al., 2000; Mojica and Diez-Villasenor, 2005).

Cas proteins – enzymes responsible for identifying and cleaving DNA.

Transcription of the CRISPR locus produces pre-CRISPR RNA (pre-crRNA), which is later processed into mature CRISPR RNA (crRNA). The crRNA guides Cas proteins to the complementary site on viral or plasmid DNA and cleaves it, thereby protecting the bacterium.

CRISPR-Cas9, also known as a type II CRISPR-Cas system, has been extensively studied due to its simplicity and flexibility. In this system, crRNA and tracrRNA (trans-activating CRISPR RNA) combine to form a single guide RNA (sgRNA). This sgRNA directs the Cas9 nuclease to the target site on DNA, where Cas9 makes a precise cut. As a result, insertions or deletions occur in the DNA sequence, disrupting the function of the targeted gene.

Because of its simplicity, efficiency, and low cost, CRISPR technology is being widely studied in almost all areas of medicine. In particular, its potential in the treatment of hereditary diseases has attracted great interest.

The CRISPR–Cas9 system consists of two main components:

Cas9 protein – a “molecular scissor” that cuts DNA at a precise location.

Guide RNA (gRNA) – a molecule that directs the Cas9 protein to the required gene site.

This system can find the target gene, cut it, and, if necessary, replace it with a new DNA fragment. In this way, genetic defects can be corrected.

Therefore, the system offers great potential for the treatment of hereditary diseases.

In sickle cell anemia, clinical studies are being conducted to correct mutations in the hemoglobin gene. In these experiments, the patient’s blood-forming stem cells are edited using CRISPR. The main goal is to deactivate the BCL11A gene, which reactivates the production of fetal hemoglobin. As a result, healthy red blood cells appear in patients.

In cancer and immunotherapy, CRISPR is used to modify T cells, giving them the ability to “recognize and destroy” cancer cells. Some clinical trials are studying the use of CRISPR in allogeneic T cells (taken from other donors).

Through this mechanism, in vivo (inside the body) gene editing is also being developed. Unlike ex vivo methods, where editing is done outside the body, in vivo editing delivers the CRISPR system directly into the patient’s body (for example, through viral vectors or lipid nanoparticles). Examples include targeting liver cells in some genetic diseases or directly editing retinal cells.

CRISPR also plays an important role in diagnostics and disease detection**.**

Conclusion.

The CRISPR–Cas9 technology is a revolutionary innovation in medical biology and genetics, providing great opportunities for treating hereditary diseases. Although it is still in the experimental and clinical trial stages, it is expected that in the near future it will become an effective treatment method for many monogenic disorders.

Thus, CRISPR technology may become an important tool in creating a healthy future for humanity free from hereditary diseases.

The CRISPR system has several advantages: it may reduce the need for constant drug injections, serve as a one-time gene therapy, correct mutations precisely, and cause fewer side effects. Moreover, the corrected mutation is not passed to the next generation.

However, there are also some limitations and risks. These include:

Off-target effects (cutting DNA in the wrong place),

Ethical issues (editing human embryos remains controversial),

Immune reactions (the body may develop an immune response against the Cas9 protein).

In summary, this system has the potential to eliminate genetic diseases completely.

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