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Applications and Ethics of Gene Editing Technology in Genetic Disease Treatment

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Abstract: Gene editing technology is fundamentally reshaping the paradigm of genetic disease treatment, and its clinical translation marks the entry of modern medicine into an era of precise intervention. The CRISPR-Cas9 system, with its programmable targeting capability, provides radical solutions for monogenic genetic diseases, achieving historic breakthroughs in the treatment of thalassemia and sickle cell disease. Technological iterations have spawned new paradigms of base editing and prime editing, enabling single-base precise correction while circumventing double-strand break risks, representing a qualitative leap in the safety dimension. Ethical controversies focus on the unpredictability of off-target effects, the intergenerational responsibility boundaries of germline editing, and the social equity of technology benefit distribution, urgently requiring the establishment of global collaborative governance mechanisms. Innovations in delivery systems have broken through organ-targeting bottlenecks, making in vivo in situ editing possible. Future development directions lie in integrating artificial intelligence to optimize editing design, perfecting lifelong safety monitoring systems, constructing ethical consensus across multicultural backgrounds, promoting deep synergy between technological innovation and humanistic care, and ultimately achieving a paradigm shift in genetic disease treatment from symptom management to etiological eradication.

Keywords: Gene Editing; Genetic Disease Treatment; CRISPR-Cas9; Ethical Governance; Off-Target Effects; Germline Editing; Base Editing; Clinical Translation

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Introduction

As a major threat to human health, genetic diseases have traditionally been treated with methods largely limited to symptom relief rather than etiological eradication. The emergence of gene editing technology brings a turning point to this dilemma, as it enables precise modification at the genomic level, fundamentally correcting pathogenic mutations[1]. The advent of CRISPR-Cas9 technology marks the entry of gene editing into the programmable era, with its operational simplicity and high efficiency rapidly making it a research hotspot. Currently, this technology has advanced from basic research to clinical application, with multiple clinical trials confirming its efficacy in treating monogenic genetic diseases. However, technological breakthroughs are accompanied by ethical challenges; permanent alterations through human germline editing trigger discussions on intergenerational responsibility, and differences in technology accessibility may exacerbate health inequalities. How to seek balance between promoting medical progress and adhering to ethical bottom lines has become a topic faced by both the scientifi c community and society. This article systematically reviews the principles of gene editing technology, current clinical application status, and ethical governance frameworks, aiming to provide references for the responsible development of this technology.

1. Basic Principles and Development History of Gene Editing Technology

1.1 Molecular Mechanisms and Core Tools of Gene Editing Technology

Gene editing technology creates double-strand breaks at specific genomic sites through sequence-specific nucleases, utilizing the cell's own repair mechanisms to achieve gene knockout, insertion, or replacement. The CRISPR-Cas9 system originates from bacterial adaptive immunity, consisting of Cas9 nuclease and single-guide RNA; the latter recognizes target sequences through base complementary pairing, guiding Cas9 to cleave DNA. Compared to zinc finger nucleases and transcription activator-like effector nucleases, CRISPR-Cas9 is simpler to design, less costly, and more efficient in editing. This system has spawned various variants, such as high-fidelity Cas9 variants that reduce off-target risks, and nickase Cas9 that achieves single-strand cleavage to reduce chromosomal translocations. Base editors fuse deaminases with Cas9 nickases, enabling C·G to T·A or A·T to G·C conversions without double-strand breaks, suitable for point mutation correction[2]. The prime editing system utilizes reverse transcriptase and engineered guide RNA, capable of executing all types of base substitutions and small fragment insertions or deletions, with editing precision reaching the single-base level.

1.2 Technological Iteration and Pathways for Editing Precision Enhancement

Gene editing technology has undergone three generations of evolution. The first-generation zinc finger nucleases achieved customized DNA recognition, but complex protein design limited their application. The second-generation TALENs utilized protein-DNA specific recognition, with improved editing efficiency but still requiring tedious assembly. The third-generation CRISPR-Cas9 technology broke through the protein design bottleneck, enabling target switching through RNA programming, revolutionarily lowering the technical threshold. Current technological optimization focuses on editing precision and safety; Cas9 protein modifications guided by structural biology have yielded high-fidelity variants SpCas9-HF1 and evoCas9, reducing off-target rates below detection limits[3]. Base editing technology has iteratively developed cytosine base editors and adenine base editors, narrowing the editing window to 1-2 nucleotides and significantly reducing bystander mutations. As fourth-generation technology, prime editing achieves precise editing through pegRNA-programmed reverse transcription templates, producing almost no double-strand break byproducts, providing new options for safe treatment.

1.3 Innovations and Challenges in Delivery Systems

Efficient and safe delivery systems are crucial for the clinical translation of gene editing. Among viral vectors, adeno-associated viruses have low immunogenicity and high tissue tropism, but packaging capacity limits their ability to carry large editing elements. Lipid nanoparticles, as non-viral vectors, encapsulate Cas9 in mRNA form and guide RNA, enabling transient expression in organs such as the liver and reducing long-term off-target risks. Virus-like particle technology combines viral efficient entry capabilities with DNA replication-free characteristics, capable of delivering ribonucleoprotein complexes that rapidly degrade after editing completion. Physical delivery methods such as electroporation are suitable for ex vivo editing of hematopoietic stem cells but are limited for in vivo applications. Current challenges lie in achieving tissue-specific targeting, blood-brain barrier penetration, and reducing innate immune responses. Novel engineered exosomes and cell-penetrating peptides provide new ideas for organ-selective delivery, with targeted ligand modifications enhancing enrichment efficiency in tumors or muscle tissues.

2. Clinical Applications of Gene Editing in Treating Monogenic Genetic Diseases

2.1 Therapeutic Breakthroughs in Hemoglobinopathies

Thalassemia and sickle cell disease, as the most common monogenic genetic diseases, have become frontier fields for the clinical translation of gene editing. The CTX001 therapy delivers CRISPR-Cas9 to autologous hematopoietic stem cells through electroporation, targeting the BCL11A erythroid enhancer to reactivate fetal hemoglobin expression, compensating for defective adult hemoglobin. Clinical trials show that transfusion-dependent β-thalassemia patients have become transfusion-independent, and sickle cell disease patients have experienced complete elimination of vaso-occlusive crises[4]. The base editing therapy BEAM-101 directly corrects the sickle cell disease mutation site, avoiding chromosomal abnormality risks associated with double-strand breaks, with early data demonstrating good safety. Gene-edited hematopoietic stem cells reconstruct normal hematopoiesis long-term after reinfusion, with single treatment potentially achieving lifelong cure, fundamentally changing disease management models.

2.2 Visual Reconstruction in Hereditary Retinal Diseases

Leber congenital amaurosis and hereditary retinal dystrophy cause irreversible vision loss, and gene editing offers possibilities for local treatment. EDIT-101 delivers CRISPR-Cas9 via subretinal injection of AAV5 vector, targeting the CEP290 gene IVS26 mutation; splice correction restores photoreceptor function. Some patients in clinical trials have shown improved dark adaptation vision and enhanced light sensitivity. Hereditary retinal diseases possess intraocular immune privilege, with local editing avoiding systemic off-target risks, and ocular structures allowing precise surgical delivery. Gene editing therapies for Stargardt disease and retinitis pigmentosa are in preclinical stages, with ABCA4 and RHO gene mutation correction strategies showing visual function protection in animal models. The combination of gene editing and optogenetics provides new pathways for visual signal reconstruction in patients with advanced retinal degeneration.

2.3 Intervention Exploration in Neurological Genetic Diseases

For neurodegenerative diseases such as Huntington's disease and amyotrophic lateral sclerosis, gene editing demonstrates disease-modifying potential. CRISPR-Cas9 targeting the HTT gene mutation region can selectively knock out mutant alleles while preserving normal gene function, with mouse models showing delayed motor symptoms. Spinal muscular atrophy promotes full-length SMN protein expression by editing the SMN2 gene splice site, with AAV9 vector-mediated intrathecal injection achieving widespread spinal motor neuron transduction. Delivery barriers are the main bottleneck; the blood-brain barrier limits systemic administration efficiency, and direct intracranial injection carries surgical risks. Focused ultrasound blood-brain barrier opening technology assists delivery, improving editing efficiency while reducing invasive procedures. Neurodegenerative diseases progress slowly, requiring long-term follow-up to assess editing persistence and safety.

3. Safety Assessment and Risk Management of Gene Editing Technology

3.1 Detection and Mitigation Strategies for Off-Target Effects

Off-target cleavage is the primary safety hazard of gene editing, potentially triggering proto-oncogene activation or tumor suppressor gene inactivation. Whole-genome off-target detection technologies include GUIDE-seq, DISCOVER-seq, and CIRCLE-seq, comprehensively assessing potential risk sites by capturing off-target break sites or detecting nuclease activity in vitro. High-fidelity Cas9 variants reduce non-specific DNA binding by mutating key amino acid residues, with SpCas9-NG and xCas9 expanding recognition ranges while enhancing specificity. Base editing and prime editing avoid double-strand breaks, fundamentally eliminating chromosomal translocation and large fragment deletion risks. Off-target effects are cell-type dependent; primary cell and stem cell editing require personalized assessment. Long-term follow-up studies show no tumor occurrence in clinical-grade edited cells, but lifelong monitoring systems need to be established.

3.2 Chromosomal Abnormalities and Genome Stability Maintenance

Double-strand break repair may trigger chromosomal translocations, inversions, and large-scale deletions, with genomic instability threatening cell survival. Microhomology-mediated end joining repair leads to unpredictable insertions or deletions, affecting gene reading frames. Single-base editing produces bystander mutations, with unintended changes in adjacent bases potentially creating new pathogenic variants. DNA damage response pathway activation triggers cell cycle arrest or apoptosis; stem cell editing requires maintaining self-renewal and multidirectional differentiation capabilities. Single-cell sequencing technology resolves edited cell clonal heterogeneity, identifying potential abnormal karyotypes. Repair pathway regulation strategies such as DNA-PK inhibitors promote homologous recombination repair, enhancing precise editing efficiency. Genome integrity assessment requires combining karyotype analysis, whole-genome sequencing, and transcriptome monitoring, ensuring editing safety through multiple dimensions.

3.3 Immunogenicity and Long-term Biological Safety

Cas9 protein originates from bacteria, and pre-existing immune memory in humans may trigger inflammatory responses, affecting editing efficiency and safety. Humoral immunity produces anti-Cas9 antibodies, and cytotoxic T lymphocytes clear edited cells. Viral vector delivery activates innate immune pathways, with interferon responses limiting transgene expression.

Non-viral delivery systems such as ribonucleoprotein complexes enable transient expression, reducing immune exposure time. Individualized immune status assessment guides delivery strategy selection, with immunosuppression protocols assisting in enhancing editing persistence. Long-term biological safety focuses on the clonal evolution of edited cells; polyclonal editing avoids single mutant cell advantage expansion[5]. Tumor risk monitoring requires integrating circulating tumor DNA detection and imaging follow-up, establishing early warning mechanisms.

4. Ethical Controversies and Social Impacts of Gene Editing Technology

4.1 Intergenerational Ethical Dilemmas of Germline Editing

Germline editing modifies genetic material that can be vertically transmitted to offspring, altering human gene pool composition and triggering controversies over intergenerational responsibility and consent rights. Heritable genome editing crosses the boundary between treatment and enhancement, with non-medical trait modification opening risks of human self-design. The international scientific community has reached consensus that current technological maturity is insufficient to support clinical application; off-target effects are amplified in embryonic development and cannot be remedied. Some countries have legislated to prohibit germline editing, but regulatory differences lead to regulatory arbitrage and cross-border reproductive tourism. Ethical frameworks need to distinguish between somatic cell therapy and germline enhancement, establishing global moratorium mechanisms until safety standards and social consensus are formed. Intergenerational justice requires contemporary people to exercise the power to alter future generations' genomes prudently, avoiding irreversible genetic consequences.

4.2 Technology Accessibility and Health Equity Challenges

The high R&D costs and complex production processes of gene editing therapies may limit their global accessibility, exacerbating inequalities in medical resource distribution. Developed countries monopolize core patents and technology platforms, making it difficult for patients in low- and middle-income countries to benefit. Personalized autologous cell therapy requires establishing GMP-grade cell processing facilities, which are difficult to implement in areas with weak medical infrastructure. Payment capacity differences make gene editing a privilege medicine, with rare disease patients facing treatment inaccessibility. Intellectual property systems and drug pricing strategies need to balance innovation incentives with global health needs; tiered pricing and voluntary licensing mechanisms enhance technology accessibility. International cooperation mechanisms promote technology transfer and capacity building, ensuring that gene editing benefits are shared inclusively.

4.3 Human Enhancement and Technology Misuse Risks

Gene editing technology applied beyond the treatment scope to cognitive enhancement, athletic performance improvement, or lifespan extension challenges medical purpose and human diversity values. Enhancement applications blur the boundaries between disease and normal variation, with social pressures driving non-medical use demands, triggering issues of compulsory enhancement and discrimination. Gene selection technology combined with editing means may lead to new eugenics practices, with specific genetic characteristics being systematically eliminated. Gene doping detection is difficult in sports competition, undermining fair competition principles. Legal regulation lags behind technological development, requiring prospective legislation to prohibit non-medical enhancement applications while protecting research freedom and medical innovation. Social dialogue mechanisms promote integration of diverse values, forming ethical consensus and normative constraints for technological development.

5. Governance Framework and Future Prospects of Gene Editing Technology

5.1 Construction and Coordination of International Regulatory Systems

Gene editing technology governance requires coordination mechanisms across national boundaries to avoid regulatory fragmentation hindering responsible innovation. The World Health Organization has established a global governance framework registry system, tracking somatic and germline clinical trials. The International Commission on the Clinical Use of Human Germline Genome Editing proposes a translation pathway for heritable editing, setting strict preconditions and moratorium periods. National regulatory systems vary significantly; the US FDA manages based on risk classification, EU advanced therapy medicinal products regulations emphasize quality and traceability, and China implements a dual-track system of ethical review and technical regulation. International coordination mechanisms promote standard mutual recognition and information sharing, preventing regulatory arbitrage while respecting cultural diversity. Soft law mechanisms such as scientific community self-discipline guidelines supplement hard law constraints, dynamically adapting to technological evolution.

5.2 Public Participation and Science Communication Mechanisms

Gene editing technology has profound social impacts, requiring inclusive public participation mechanisms to integrate diverse stakeholder voices. Citizen juries and consensus conference models promote in-depth understanding of technical risks and ethical dimensions among lay publics, transcending simple support or opposition binary thinking. Science communication strategies need to avoid technological determinism narratives, presenting uncertainty and developmental openness to build public trust. Patient communities participate in research priority setting and clinical trial design, ensuring therapy development aligns with real needs. Media responsibility lies in accurately reporting technological breakthroughs and risks, avoiding excessive hype or fear-mongering. Education system construction enhances public scientific literacy, cultivating critical thinking skills to support democratic decision-making processes.

5.3 Technological Evolution and Clinical Translation Prospects

Gene editing technology continues to develop toward precision and intelligence; artificial intelligence-assisted guide RNA design enhances targeting specificity, predicting off-target sites and editing efficiency. In vivo direct editing technology breaks through the limitations of ex vivo cell processing, with lipid nanoparticles and virus-like particles achieving organ-selective delivery. Multiplex editing technology simultaneously corrects multiple gene sites, applicable to polygenic genetic diseases and complex diseases. Automation and closed manufacturing systems reduce production costs and enhance therapy accessibility. Clinical translation accelerates, with indications expanding from monogenic genetic diseases to common chronic diseases; gene editing therapies for cardiovascular diseases and metabolic diseases enter early clinical stages. Long-term safety database establishment and real-world evidence accumulation improve risk-benefit assessment systems.

6. Conclusion

Gene editing technology opens revolutionary pathways for genetic disease treatment, advancing from proof-of-concept to clinical practice, demonstrating potential to cure previously incurable diseases. Technological iterations continuously optimize editing precision and safety; base editing and prime editing reduce off-target risks, and novel delivery systems expand treatment scope. However, technological breakthroughs are accompanied by profound ethical challenges regarding the intergenerational impact of germline editing, technology accessibility inequality, and the boundaries of human enhancement, requiring responses from both the scientific community and society. Establishing inclusive governance frameworks that integrate strict safety supervision, ethical review, and public participation is the guarantee for responsible technological development. Future development directions lie in deepening basic mechanism research, perfecting long-term safety assessment, promoting global fair accessibility, and ensuring that gene editing technology truly serves human health and well-being. In the tension between scientific exploration and ethical reflection, a cautiously optimistic attitude will guide this technology toward a tomorrow that benefits humanity, achieving harmonious unity between precision medicine and humanistic values.

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Conflict of Interests

The authors declare that there is no conflict of interest regarding the publication of this paper.

Reference

[1] Zhang, Q., & Gu, M. L. (2022). Gene editing technology and its clinical application. Chemistry of Life, 42(01), 41–55.

[2] Sun, X. F. (2021, August 3). Construction and key technology research and development of monogenic genetic disease

iPS cell bank and its efficient gene editing platform [Unpublished project report]. The Third AffiliatedHospital of Guangzhou Medical University, Guangdong Province.

[3] Qiu, Z. C., Li, Z. F., & Shi, H. (2021). Gene editing technology and its research progress and application prospects in disease treatment. Journal of Kunming University of Science and Technology (Natural Science Edition), 46(05), 100–109.

[4] Anonymous. (2020). Gene editing technology successfully treats genetic disease for the first time. Chinese Journal of Clinical Oncology and Rehabilitation, 27(08), 1024.

[5] Xie, F. Y. (2019). Application progress of gene editing technology in gene therapy. Biological Chemical Engineering, 5(01), 162–164.