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Request: Advances in CRISPR-Cas gene editing and implications for rare genetic diseases

Summary

Advances in CRISPR-Cas gene editing have significantly impacted the treatment of rare genetic diseases, offering new therapeutic possibilities and hope for patients. The paper "Current progress in CRISPR-Cas systems for rare diseases" highlights the profound influence of CRISPR-Cas technology on molecular biology and genetics, emphasizing its role in gene function research, animal disease models, and patient genetic therapy. This technology allows for precise alterations in the genetic code, which is crucial for developing treatments for rare diseases. "In vivo prime editing rescues photoreceptor degeneration in nonsense mutant retinitis pigmentosa" discusses the application of prime editing, a next-generation gene editing tool, to correct point mutations with high efficiency and minimal off-target effects. This study demonstrates the potential of prime editing in treating hereditary diseases like retinitis pigmentosa, a common inherited retinal dystrophy. The article "Novel CRISPR-Cas9 iPSC knockouts for PCCA and PCCB genes: advancing propionic acidemia research" presents the creation of induced pluripotent stem cell (iPSC) lines with CRISPR-Cas9 knockouts for the PCCA and PCCB genes. This advancement provides a robust platform for studying propionic acidemia, a rare metabolic disorder, and developing therapeutic interventions. "Challenges and progress related to gene editing in rare skin diseases"

explores the application of CRISPR-based technologies in treating genodermatoses, a group of inherited skin disorders. The paper highlights the potential of these technologies to offer promising therapeutic prospects, addressing the clinical manifestations and secondary complications associated with these conditions. Lastly, "Rare genetic disorders in India: Current status, challenges, and CRISPR-based therapy" discusses the significant impact of CRISPR-Cas9 technology on genetic therapies for rare diseases in India. The emergence of CRISPR-based treatments provides new hope for individuals affected by these life-threatening disorders, which often result from single-gene mutations. Together, these papers underscore the transformative potential of CRISPR-Cas and related gene editing technologies in advancing the treatment of rare genetic diseases, paving the way for personalized and effective therapeutic strategies.

Relevant Articles

1. Genomic medicine and personalized treatment: a narrative review.

Date: 2025-02-13

Source: PubMed

DOI: 10.1097/MS9.0000000000002965

URL: https://pubmed.ncbi.nlm.nih.gov/40213198

Abstract: Genomic medicine, which integrates genomics and bioinformatics into clinical care and diagnostics, is transforming healthcare by enabling personalized treatment approaches. Advances in technologies such as DNA sequencing, proteomics, and computational power

have laid the foundation for individualized therapies that account for genetic variations influencing disease risk, progression, and treatment response. This review explores the historical milestones leading to current applications of genomic...

2. In vivo prime editing rescues photoreceptor degeneration in nonsense mutant retinitis pigmentosa.

Date: 2025-03-10

Source: PubMed

DOI: 10.1038/s41467-025-57628-6

URL: https://pubmed.ncbi.nlm.nih.gov/40064881

Abstract: The next-generation gene editing tool, prime editing (PE), is adept at correcting point mutations precisely with high editing efficiency and rare off-target events and shows promising therapeutic value in treating hereditary diseases. Retinitis pigmentosa (RP) is the most common type of inherited retinal dystrophy and is characterized by progressive degeneration of retinal photoreceptors and, consequently, visual decline. To date, effective treatments for RP are lacking. Herein, a PE system is d...

3. Novel CRISPR-Cas9 iPSC knockouts for PCCA and PCCB genes: advancing propionic acidemia research.

Date: 2025-03-05

Source: PubMed

DOI: 10.1007/s13577-025-01193-z

URL: https://pubmed.ncbi.nlm.nih.gov/40044943

Abstract: Propionic acidemia (PA) is a rare autosomal recessive metabolic disorder caused by mutations in the PCCA and PCCB genes, which encode subunits of the mitochondrial enzyme propionyl-CoA carboxylase (PCC). This enzyme deficiency leads to the accumulation of toxic metabolites, resulting in severe metabolic dysfunction. To create ideal in vitro disease models of PA with isogenic controls and provide a robust platform for therapeutic research, we generated two induced pluripotent stem cell (iPSC) lin...

4. Current progress in CRISPR-Cas systems for rare diseases.

Date: 2024-08-31

Source: PubMed

DOI: 10.1016/bs.pmbts.2024.07.019

URL: https://pubmed.ncbi.nlm.nih.gov/39824580

Abstract: The groundbreaking CRISPR-Cas gene editing method permits exact genetic code alteration. The

"CRISPR" DNA protects bacteria from viruses. CRISPR-Cas utilizes a guide RNA to steer the Cas enzyme to the genome's gene editing target. After attaching to a sequence, Cas enzymes cleave DNA to insert, delete, or modify genes. The influence of CRISPR-Cas technology on molecular biology and genetics is profound. It allows for gene function research, animal disease models, and patient genetic therapy. Gen...

5. Reporter Mice for Gene Editing: A Key Tool for Advancing Gene Therapy of Rare Diseases.

Date: 2024-09-09

Source: PubMed

DOI: 10.3390/cells13171508

URL: https://pubmed.ncbi.nlm.nih.gov/39273078

Abstract: Most rare diseases are caused by mutations and can have devastating consequences. Precise gene editing by CRISPR/Cas is an exciting possibility for helping these patients, if no irreversible developmental defects have occurred. To optimize gene editing therapy, reporter mice for gene editing have been generated which, by expression of reporter genes, indicate the efficiency of precise and imprecise gene editing. These mice are important tools for testing and comparing novel gene editing methodol...

6. Gene editing in liver diseases.

Date: 2024-07-30

Source: PubMed

DOI: 10.1002/1873-3468.14989

URL: https://pubmed.ncbi.nlm.nih.gov/39079936

Abstract: The deliberate and precise modification of the host genome using engineered nucleases represents a groundbreaking advancement in modern medicine. Several clinical trials employing these approaches to address metabolic liver disorders have been initiated, with recent remarkable outcomes observed in patients with transthyretin amyloidosis, highlighting the potential of these therapies. Recent technological improvements, particularly CRISPR Cas9-based technology, have revolutionized gene editing, e...

7. Towards a Cure for Diamond-Blackfan Anemia: Views on Gene Therapy.

Date: 2024-05-27

Source: PubMed

DOI: 10.3390/cells13110920

URL: https://pubmed.ncbi.nlm.nih.gov/38891052

Abstract: Diamond-Blackfan anemia (DBA) is a rare genetic disorder affecting the bone marrow's ability to produce red blood cells, leading to severe anemia and various physical abnormalities. Approximately 75% of DBA cases involve heterozygous mutations in ribosomal protein (RP) genes, classifying it as a ribosomopathy, with RPS19 being the most frequently mutated gene. Non-RP mutations, such as

in GATA1, have also been identified. Current treatments include glucocorticosteroids, blood transfusions, and h...

8. Challenges and progress related to gene editing in rare skin diseases.

Date: 2024-03-23

Source: PubMed

DOI: 10.1016/j.addr.2024.115294

URL: https://pubmed.ncbi.nlm.nih.gov/38527624

Abstract: Genodermatoses represent a large group of inherited skin disorders encompassing clinically-heterogeneous conditions that manifest in the skin and other organs. Depending on disease variant, associated clinical manifestations and secondary complications can severely impact patients' quality of life and currently available treatments are transient and not curative. Multiple emerging approaches using CRISPR-based technologies offer promising prospects for therapy. Here, we explore current advances ...

9. Rare genetic disorders in India: Current status, challenges, and CRISPR-based therapy.

Date: 2024-02-01

Source: PubMed

URL: https://pubmed.ncbi.nlm.nih.gov/38383973

Abstract: Rare genetic diseases are a group of life-threatening disorders affecting significant populations worldwide and posing substantial challenges to healthcare systems globally. India, with its vast population, is also no exception. The country harbors millions of individuals affected by these fatal disorders, which often result from mutations in a single gene. The emergence of CRISPR-Cas9 technology, however, has ushered in a new era of hope in genetic therapies. CRISPR-based treatments hold the po...

10. Next-generation therapeutics for rare genetic disorders.

Date: 2024-04-01

Source: PubMed

DOI: 10.1093/mutage/geae002

URL: https://pubmed.ncbi.nlm.nih.gov/38332115

Abstract: The therapeutic potential of the human genome has been explored through the development of next-generation therapeutics, which have had a high impact on treating genetic disorders. Classical treatments have traditionally focused on common diseases that require repeated treatments. However, with the recent advancements in the development of nucleic acids, utilizing DNA and RNA to modify or correct gene expression in genetic disorders, there has been a paradigm shift in the treatment of rare disea...

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