



## Advances in Gene Therapy for Rare Diseases: Clinical Trials and the Directions

Mia Linwood\*

Department of Genomes and Proteomics, Peking University, Beijing, China

### DESCRIPTION

Gene therapy, a promising frontier in medical science, has seen significant advancements over the past decade. This innovative approach involves the modification of a patient's genetic material to treat or prevent disease. Rare diseases, often caused by genetic mutations, are prime candidates for gene therapy. With over 7,000 identified rare diseases affecting millions worldwide, gene therapy for many conditions that currently lack effective treatments. Recent clinical trials have demonstrated the significant of gene therapy, and directions point towards even more transformative.

#### Clinical trials: Pioneering successes

The journey of gene therapy from experimental concept to clinical reality has been marked by several landmark trials. One of the most notable successes is the treatment of Spinal Muscular Atrophy (SMA), a severe genetic disorder that leads to muscle wasting and early death. The approval of onasemnogene abeparvovec (Zolgensma) marked a significant milestone. This gene therapy delivers a functional copy of the Survival Motor Neuron-1 (SMN1) gene using an Adeno-Associated Virus (AAV) vector, showing remarkable efficacy in improving motor function and survival rates in infants with SMA.

Similarly, Luxturna (voretigene neparvovec) became the first Food and Drug Administration (FDA)-approved gene therapy for an inherited retinal disease, specifically targeting Leber Congenital Amaurosis (LCA), a condition leading to blindness. Luxturna works by delivering a normal copy of the *RPE65* gene directly into retinal cells, restoring vision in patients who previously had no treatment options.

Hemophilia, a rare bleeding disorder, has also seen promising results from gene therapy. Trials involving the infusion of a functional gene to produce clotting factors have demonstrated sustained increases in clotting factor levels, reducing the need for regular infusions and significantly improving patients' quality of life.

#### Challenges and innovations

Despite these successes, gene therapy faces several challenges. One major hurdle is the delivery of therapeutic genes to target cells without eliciting an immune response. Viral vectors, while effective, can sometimes provoke immune reactions or have limited capacity for large genes. To address this, researchers are exploring non-viral delivery methods, such as lipid nanoparticles and CRISPR-Cas9 systems, which offer more precise gene editing capabilities and reduced immunogenicity.

Another challenge is ensuring long-term expression and stability of the introduced genes. Integrating vectors, like lentiviruses, significant solution by incorporating the therapeutic gene into the host genome, providing permanent correction. However, this approach carries the risk of insertional mutagenesis, which could significantly activate oncogenes and lead to cancer. Balancing efficacy and safety remains a critical focus of ongoing research.

#### Directions: Towards precision medicine

The gene therapy for rare diseases lies in the continued refinement of these technologies and the expansion of their applications. Personalized gene therapy, customized to an individual's unique genetic makeup, represents a significant advancement. By leveraging Next-Generation Sequencing (NGS) and bioinformatics, scientists can identify specific mutations responsible for a patient's condition and design custom therapies that target those mutations with high precision.

Emerging technologies like base editing and prime editing capable immense promise for the gene therapy. These techniques enable precise alterations to the DNA sequence without causing double-strand breaks, minimizing the risk of unintended genetic changes. This precision opens up possibilities for treating a broader range of genetic disorders with greater accuracy and safety.

Advances in gene therapy have brought new perspective to individuals suffering from rare genetic diseases. Clinical trials have demonstrated this innovative approach to provide lasting

**Correspondence to:** Mia Linwood, Department of Genomes and Proteomics, Peking University, Beijing, China, E-mail: linwood@gmail.com

**Received:** 03-Jun-2024, Manuscript No. BOM-24-26365; **Editor assigned:** 05-Jun-2024, Pre QC No. BOM-24-26365 (PQ); **Reviewed:** 19-Jun-2024, QC No. BOM-24-26365; **Revised:** 26-Jun-2024, Manuscript No. BOM-24-26365 (R); **Published:** 03-Jul-2024, DOI: 10.35248/2167-7956.24.13.388

**Citation:** Linwood M (2024) Advances in Gene Therapy for Rare Diseases: Clinical Trials and the Directions. J Biomol Res. 13:388.

**Copyright:** © 2024 Linwood M. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

and transformative treatments. While challenges remain, ongoing research and technological advancements for safer, more effective, and personalized therapies. The gene therapy is

bright, to deliver that will change the lives of countless patients and redefine the landscape of rare disease treatment.