**Q: How is Pfizer Rare Disease advancing research in gene therapy?**

A: Pfizer Rare Disease is advancing research on gene therapy as one of the next developments in delivering potentially transformational medicines to people living with genetic diseases. With our commitment to rare disease patients, we look to gene therapy as an opportunity to improve the lives of people who have complex diseases with significant unmet needs.

**Q: What is Pfizer’s focus in gene therapy?**

A: Currently, Pfizer Rare Disease is focused on gene therapy treatments in diseases that have single gene defects, such as certain neuromuscular and hematologic diseases.

Our approach is highly specialized, potentially one-time gene therapy treatments that use custom-made vectors modeled after the Adeno-Associated Virus (AAV) to potentially deliver treatment effectively to patients. It is a technology that can be standardized, streamlining the manufacturing and regulatory path to medicine approval.

**Q: What is a genetic disease?**

A: A genetic disease is the result of changes, or alterations, in a person’s DNA, some of which can be debilitating and life-threatening. This can occur either when a person inherits a nonfunctioning gene, or in rare cases, with a spontaneous alteration. Genes play an essential role in determining the function of each cell in the body, making up 30 million codes of DNA. If even one of these codes is damaged, a genetic alteration may occur causing a genetic disease.

**Q: What is gene therapy?**

A: Gene therapy seeks to deliver functioning genes in the body, allowing a person to produce the necessary protein they were unable to make on their own. This process is a potentially one-time treatment that uses a vector—often, a modified virus with no viral DNA present—as a custom-made vehicle that delivers the functioning gene to a specific targeted tissue.

**Q: How long does gene therapy last?**

A: While gene therapy holds promise for people with genetic diseases, it will not be an appropriate solution for every patient. Clinical trials are currently underway to explore the many unknowns, including how long the therapy will last. That said, evidence to date indicates gene therapy has the potential to restore normal function in affected tissues or cells over the long-term that may enable a patient to manage his or her disease without the need for ongoing treatments.

**Q: What does gene therapy do to an individual’s DNA?**

A: Pfizer’s current approach to gene therapy does not alter a person’s DNA to be effective, also known as nonintegrating gene therapy. This means that our approach has a very low likelihood of affecting an individual cell’s genetic material. Because of this, the existing genetic alteration can still be inherited from parent to child. Our approach is also classified as in vivo. With in vivo gene therapy, the genes are introduced directly into a person’s body.

**Q: What are the potential benefits of gene therapy?**

A: While gene therapy holds promise for people with genetic diseases, it will not be an appropriate solution for every patient. Gene therapy is a treatment option with life-changing potential. It is a potentially one-time treatment that could enable a patient to manage their disease without the need for ongoing treatment, and allow more life choices so a person can live a more “normal” life.
What are the potential challenges of gene therapy?

Factors that may make someone ineligible to receive gene therapy treatment include patients with pre-existing antibodies that would neutralize the specific gene therapy treatment, patients who have previously received gene therapy and developed these antibodies, and for certain disease trials, patients who are not yet adults. Eligibility will be explored with a blood test to check for antibodies to the custom vector, and patients can discuss the results with their physicians and determine how to proceed on an individual basis.

How do you know if you are eligible for gene therapy?

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What are antibodies?

Antibodies work with the immune system to identify and neutralize foreign objects, such as bacteria, viruses, or certain proteins. While the AAV vector contains no viral DNA, the capsule or shell remains and may provoke an immune response, such as the development of antibodies.

How has gene therapy evolved since the 1990s?

The historical safety and efficacy issues led to scientific advancements in gene therapy, resulting in evolution of the technology. Direct outputs include the development of safer vectors for DNA delivery, and improved study protocols and patient consent information.

What are other approaches to genetic medicine currently being explored?

Gene editing is another potential treatment for genetic diseases currently being explored, in which the patient’s chromosomal DNA is directly altered to correct a genetic error.

How do you manufacture a gene therapy treatment?

Pfizer Rare Disease will manufacture gene therapy treatments using production processes that are very similar to other biotechnology products such as monoclonal antibodies and vaccines. The process uses recombinant cell culture technology and purification followed by sterile vial filling. The similarity to traditional biotech processes means higher confidence on the scale up, reproducibility, and the design of manufacturing facilities.

When will gene therapy be available?

Pfizer Rare Disease is actively researching gene therapy and various clinical trials are underway. It is still too early to provide a time frame as to when gene therapy will be approved for use, but we are hopeful for the future.

Where is Pfizer Rare Disease in the process in regards to gene therapy?

Clinical trials conducted by Pfizer are currently underway, and we are committed to offering this transformative medicine as soon as possible. Patients can learn more about our clinical trials through our company’s “Find a Trial” page on our Website or through clinicaltrials.gov.

The health information contained herein is provided for educational purposes only and is not intended to replace discussions with a health care provider. All decisions regarding patient care must be made with a health care provider, considering the unique characteristics of the patient.