

Genomic Medicine: Unique Considerations for Rare Disease Clinical Trials



Genomic medicine is an emerging field of study that looks at one's genes to determine how errors or changes in an individual's DNA may result in disease and uses that knowledge to create a treatment to correct the error. Gene therapy, genome editing, gene-edited cell therapy and genome regulation are all types of genomic medicine that work inside the body's cells to address errors in DNA or changes in a person's genes that can result in genetic and inherited disorders.

There is great optimism among genomic medicine researchers that someday soon a person with a disease like hemophilia, who currently receives ongoing treatments, may be able to receive a one-time single dose therapy that is designed to work from inside the body's cells to stop or slow the progression of disease. This type of treatment addresses the underlying cause of the disorder, helping the body better fight disease by tackling it at the source. If successful, this person would be able to stop long-term and constant treatment for hemophilia.

Any drug or treatment must move through several phases of testing called clinical trials to assess safety and effectiveness before it can be approved by a regulatory agency such as the U.S. Food and Drug Administration (FDA) or the European Medicines Agency (EMA) for use by patients.

Regardless of the treatment being studied, there are certain components that are similar for all clinical trials. For example, clinical trials are conducted to determine if a treatment works without too many side effects. There are strict guidelines about who qualifies to participate in a study. Clinical trials for genomic medicines always involve patient volunteers and are highly regulated to protect the patients' rights and safety.

Clinical trials may have unique considerations when the treatment being considered is a genomic medicine. Because the intent of a genomic medicine is to provide a one-time treatment with long-term therapeutic effects, there are some differences in the way these therapies are developed and investigated in clinical trial patients as opposed to a medicine that can be stopped at any time or doesn't have long-term effects.



Unique Considerations for Genomic Medicine Clinical Trials

There are a variety of tests that could be performed before or during the clinical trial. If the disease is caused by a genetic defect, researchers could perform genetic testing on a patient volunteer to ensure correct diagnosis of the disorder they are studying.

Researchers may also conduct testing to see if a person's immune system has existing antibodies that could block the treatment before it is able to reach the body's cells. A person would be ineligible for a clinical trial if they had immunity to any part of the treatment.

When clinical trials reach the phase where volunteers are needed, patients with all stages of disease progression may be recruited for participation. For genomic medicine clinical trials, researchers may seek patient volunteers who are at less severe stages of the disease. This is because as disease symptoms worsen

over time, a researcher may not be able to tell the difference from progressive challenges of living with the specific condition or side effects from the medicine being tested.

Generally, in non-genomic medicine clinical trials, healthy volunteers may be included in the early phases of a study. However, due to the long-acting nature of genomic medicine and the types of rare diseases they seek to treat, healthy volunteers are not usually included in these studies.

Clinical trials typically move through three phases, enrolling hundreds or thousands of volunteers as the phases progress. Rare or ultra-rare diseases with very limited patient populations may move through development with a smaller number of patients or combine the clinical trial phases.

With the goal of genomic medicine to become a long-term treatment, it becomes critical to follow the individual over an extended period to monitor and assess the safety of the treatment and impact on their health. Unlike patients in a clinical trial where the treatment leaves the body quickly, patients who receive a genomic medicine treatment could expect researchers to follow them for 10-15 years, or even longer.

One additional consideration is the informed consent process. Informed consent is an ongoing interactive discussion to help patients make decisions about whether to begin or continue to participate in a clinical trial. This is a critical ethical consideration in every clinical trial and the process helps a potential volunteer understand what to expect during a trial, procedures and tests that will be conducted and potential benefits and risks. With the long-term nature of genomic medicine, the study team should proactively provide education about the trial and participants should be empowered to ask questions to fully understand the commitment, the benefits and the risks before deciding whether to participate in a research study.



Understanding the Patient Experience with Rare Diseases

Researchers also review other kinds of information about a patient's personal experience with a disease to understand illness trends, treatment outcomes, and some demographics like age and gender. Often, this information helps researchers make decisions about how clinical trials are designed or the parameters they want to measure during the trial.

Learn More

For some diseases there are no treatments at all. The excitement about certain genomic medicines is the possibility of long-term effect without the need for ongoing interventions. However, without the patients who volunteer to participate in clinical trials or share their experience with a disease or condition, the development of new medicines would not be possible.

Consider connecting with a patient advocacy group to understand clinical trial opportunities or to participate in patient experience data collection. Find rare disease organizations by searching the resources at any of the following databases:

- The [National Organization for Rare Disorders \(NORD\) Organizational Database](#) provides a comprehensive list of organizations to help patients and families affected by rare diseases.
- The [Global Genes Rare List](#) is a database with information on specific diseases, support organizations, related news, events and clinical trials.
- In partnership with [Genetic Alliance](#), [Disease InfoSearch](#) connects individuals to support organizations and avenues to participate in research.