Clinical Research in rare genetic disorders

Clinical research is medical research that in this scenario, involves individuals with rare genetic disorders to evaluate safety and efficacy of new treatments and therapies.

Clinical Trial

A clinical research study in which one or more human subjects are prospectively assigned to one or more interventions (which may include placebo or other control groups) to evaluate the effects of those interventions on health-related medical, developmental or behavioral outcomes.

Healthy Volunteer

A healthy volunteer is a person with no known significant health problems who participates in a clinical research study to evaluate a new drug, device, or intervention.

Inclusion/Exclusion Criteria

Factors that allow someone to participate in a clinical trial are *inclusion criteria*. Those that exclude or not allow participation are *exclusion criteria*. Examples for *inclusion criteria* are: age (adults versus pediatric), diagnosis of a rare genetic disorder, regular vision and hearing. Examples for *exclusion criteria* include: participation in another clinical trial/clinical research, presence of clinical symptoms that may interfere with study participation among others.

Informed Consent

Informed consent explains what study tasks/interventions are involved in participation as well as risks and potential benefits about a clinical trial before someone decides whether to participate. It also formally asks a patient/guardian to enroll into the clinical trial.

Patient participant

A patient participant has a rare genetic condition and participates in research to better understand, diagnose, treat, or cure that condition.

Phases of Clinical Trials

Clinical trials are conducted in “phases.” The trials at each phase have a different purpose and help researchers answer different questions.

- **Phase I trials** — An experimental drug or treatment is given to a small group of people (20–80) for the first time. The purpose is to evaluate its safety and identify side effects.
- **Phase II trials** — The experimental drug or treatment is administered to a larger group of people (100–300) to determine its effectiveness and to further evaluate its safety.
- **Phase III trials** — The experimental drug or treatment is administered to large groups of people (1,000–3,000) to confirm its effectiveness, monitor side effects, compare it with standard or equivalent treatments.
- **Phase IV trials** — After a drug is licensed and approved by the FDA researchers track its safety, seeking more information about its risks, benefits, and optimal use.

Placebo

A placebo is a pill or liquid that looks like the new treatment but does not have any treatment value from active ingredients. It serves as a control to determine the safety and efficacy of the treatment.

Protocol

A protocol is a carefully designed plan to safeguard the participants' health and answer specific research questions.

Principal Investigator

A Principal Investigator is a doctor who leads the clinical research team and, along with the other members of the research team, regularly monitors study participants' health to determine the study's safety and effectiveness. The Principal Investigator is responsible for the conduct of the clinical trial.

Randomization

Randomization is the process by which two or more alternative treatments are assigned to study participants by chance rather than by choice.

Single- or Double-Blind Studies

Single-blind (also called single -masked studies) are studies in which the study participants do not know which medicine is being used, so they can describe what happens without bias.

Double-blind studies (also called double-masked studies) are studies in which both the study participants and the study researchers do not know which medicine is being used. This allows participants to describe what happens without bias and researchers to prevent bias in interpretating results.

Types of Clinical Trials

- **Diagnostic trials** determine better tests or procedures for diagnosing a particular disease or condition.
- **Natural history studies** provide valuable information about how genetic disorders progress or change over time.
- **Quality of life trials** (or supportive care trials) explore and measure ways to improve the comfort and quality of life of people with a genetic disorder.
- **Screening trials** test the best way to detect certain diseases or health conditions.
- **Treatment trials** test new treatments, new combinations of drugs, or new approaches to surgery or radiation therapy.

Adapted from

https://www.nih.gov/health-information/nih-clinical-research-trials-you/glossary-common-terms