Achondroplasia — the most common form of human dwarfism — is caused by a specific mutation in the gene that produces the FGFR3 protein.

The FGFR3 gene normally instructs the body to stop producing FGFR3 when certain parts of the body fully developed. In people with achondroplasia, the FGFR3 gene is turned on to make FGFR3 all the time. This causes the bones of the body to keep growing even after they should have stopped growing.

The result is disproportionate short stature. People with achondroplasia may have a small head in comparison to their bodies. They may also have a small chest and short limbs. People with achondroplasia also may also have problems with their spine and joint mobility, and increased risk of other conditions such as sleep apnea, ear infections, and obesity.

Achondroplasia can also lead to potentially serious complications over a person’s lifetime that are not as easy to see. These complications can impact various aspects of a person’s life, such as lower limb movement, difficulty bending over, and sleep apnea. In the United States, the average life expectancy for adults with achondroplasia is 66 years.

**Clinical Trial Process**

The goal of clinical research is to gather information about the safety and effectiveness of a new product — whether it’s a drug, a treatment, or a medical device — in people with a certain condition. There are several phases in the clinical trial process.

**Phase 1**

Typically includes a small group of people (fewer than 100) who have the condition. The purpose of the trial is to evaluate safety, determine suitable doses and schedule of administration, and understand the maximum tolerated dose.

**Phase 2**

Involves more patients — often from 100 to several hundred — as a more precise measurement of the drug’s effectiveness is required. The trial may be divided into three phases:

- The initial phase is designed to evaluate safety; the drug is tested in small groups of patients.
- The second phase is designed to determine the drug’s effectiveness; the drug is tested in larger groups of patients.
- The third phase is designed to evaluate effectiveness and dosage; the drug is tested in typical populations.

**Phase 3**

The trial is divided into three phases:

- The initial phase is designed to evaluate safety; the drug is tested in small groups of patients.
- The second phase is designed to determine the drug’s effectiveness; the drug is tested in larger groups of patients.
- The third phase is designed to evaluate effectiveness and dosage; the drug is tested in typical populations.

**Regulatory Review**

The data from clinical trials is used to prepare a regulatory application (MAA) that is submitted to regulatory authorities. The regulatory authorities review the data and decides whether or not to approve the drug.

**BMN 111**

BioMarin is in the process of working with health authorities to design the clinical trial process to demonstrate the potential of BMN 111 to help people with achondroplasia. The primary objective of BMN 111 is to evaluate safety, tolerability, and how BMN 111 may work on the underlying problem to signal regular bone growth.

**References**


**BioMarin**

Committed to Our Work in Achondroplasia

BioMarin is in the process of working with health authorities to design the clinical trial process to demonstrate the potential of BMN 111 to help people with achondroplasia. The primary objective of BMN 111 is to evaluate safety, tolerability, and how BMN 111 may work on the underlying problem to signal regular bone growth.

**Roadmap to a New Therapy**

BioMarin is committed to exploring therapies for conditions such as achondroplasia. Committed to Work in Achondroplasia and the Clinical Trial Process

**Clinical Trial**

BioMarin is in the process of working with health authorities to design the clinical trial process to demonstrate the potential of BMN 111 to help people with achondroplasia. The primary objective of BMN 111 is to evaluate safety, tolerability, and how BMN 111 may work on the underlying problem to signal regular bone growth.