

BioMarin's BMN 349 Alpha-1 Antitrypsin Deficiency Research Program: An Update for the Alpha-1 Community

On November 30, 2021 BioMarin announced intention to add Alpha-1 Antitrypsin (A1AT) Deficiency to their pipeline of conditions being researched.

BioMarin is a global pharmaceutical company with 7 approved therapies and more than 20 years of experience in developing innovative medicines for rare genetic conditions. Each investigative medicine we pursue is guided by a fundamental understanding of the genetics and underlying biology of the condition it will address. We focus on genetic conditions where we believe we can make a meaningful impact.

About the Biology of A1AT Deficiency



The underlying genetic cause of A1AT deficiency is a variation in the SERPINA1 gene. The most common deficiency-causing variant is the Z allele. A1AT protein produced from a Z allele (zA1AT) is not properly folded into its final structure in the liver cells where it is produced and isn't released into the body. As a result, this mis-folded protein builds up and aggregates within the liver cells, potentially leading to liver disease, and the level of the protein circulating in the body is lowered, potentially leading to lung disease.

About BMN 349 Research for A1AT Deficiency

BMN 349 is an investigational orally administered molecular chaperone that is being researched to understand its safety and to see if it can impact folding of the zA1AT protein, having an effect on secretion to the rest of the body. BMN349 has not been approved for use or determined to be safe or effective.

Before a new treatment can be made available, it must go through careful testing in clinical studies to see if it is safe and effective. BMN 349 has not yet begun clinical studies in humans. Pre-clinical studies, also known as animal studies, are intended to support an investigational new drug (IND) application with regulatory authorities which would allow clinical trials to begin in humans. If results from the ongoing pre-clinical studies are supportive, BioMarin anticipates filing an IND with the FDA for BMN 349 in 2023 which would allow clinical trials in humans to begin.

For additional information:



• Contact BioMarin Medical Information toll free at 1-800-983-4587 or medinfo@bmrn.com