

Audentes Therapeutics

INTRODUCTION

Rare diseases have undefined patient populations who are undiagnosed or misdiagnosed, healthcare providers who are unaware of disease states and their manifestations, and treatment journeys that are not well-understood. IPM.ai transforms real world data into real world insights that uncover the ideal patient, their treatment journey and their healthcare ecosystem so that life sciences companies can accelerate the successful development and commercialization of life-changing therapies for specialty and rare diseases that lead to optimal patient outcomes.

THE CONDITION

X-Linked Myotubular Myopathy (XLMTM), is a rare and life-threatening neuromuscular disease that is characterized by extreme muscle weakness, respiratory failure, and early death. The condition is caused by mutations in the MTM1 gene that lead to a lack or dysfunction of myotubularin, a protein that is needed for normal development, maturation, and function of skeletal muscle cells. Affecting approximately 1 in 50,000 newborn males, XLMTM, which is devastating for parents and their children, places a substantial burden on our healthcare system, including high rates of general utilization, hospitalization, and surgical intervention.

THE CHALLENGE

To address this crucial unmet market need, Audentes Therapeutics is developing a novel product candidate AT132, an AAV8 vector containing a functional copy of the MTM1 gene, for the treatment of XLMTM. To date, the company has reported promising safety, efficacy, and muscle biopsy data from ASPIRO, an ongoing, multicenter, ascending dose Phase 1/2 clinical study designed to evaluate the safety and efficacy of AT132. The therapy was granted Regenerative Medicine and Advanced Therapy (RMAT), Rare Pediatric Disease, Fast Track and Orphan Drug designations by the United States Food and Drug Administration, and Priority Medicines (PRIME)

and Orphan Drug designations by the European Medicines Agency. Until October 2020 there was no ICD 10 code, and due to the uncommon nature of XLMTM, diagnosis can often be delayed or nonexistent. Audentes Therapeutics partnered with IPM.ai to develop a machine learning model to identify undiagnosed patients and relevant health care providers as well as characterize disease burden for various patient types via artificial intelligence and medical claims data.

THE SOLUTION

IPM.ai anonymized and imported registry data from an academic Key Opinion Leader to help uncover known patients in our real world data universe of over 300 million de-identified patients. We then designed a machine learning algorithm to identify highly likely, yet currently undiagnosed patients, and developed comprehensive patient profiles across multiple patient segments to enhance disease understanding and paint a more robust picture of the patient treatment journey.

THE OUTCOME

IPM.ai constructed an ongoing, biweekly feed of likely undiagnosed patients in conjunction with alerts of new activity, providing the field with timely insights. To date, the combination of predictive and rules-driven alerts has uncovered over 10% of known XLMTM patients.

At the same time, IPM.ai and Audentes have successfully unlocked a deeper understanding of the XLMTM patient's journey across a multitude of analyses. New insights provide clarity into the patient flow through various stages of diagnoses and procedures, highlight the treatment paradigm across healthcare provider specialties, illustrate the frequent comorbidities and misdiagnoses, and depict hospitalization patterns through the care pathway. In collaboration with three leading physicians, the team is preparing a submission of these novel findings for publication.

About IPM.ai

IPM.ai, part of Real Chemistry, is an Insights as a Service (IaaS) provider that empowers the world's leading life sciences companies to better understand and improve the lives of patients through the development and commercialization of precision medicine for specialty and rare diseases. IPM's system of insight optimizes drug development, clinical study, product launch and commercial operations through granular-level longitudinal analytics, artificial intelligence and machine learning in conjunction with a real world data universe of over 300 million de-identified patients and 65 billion anonymized social determinants of health signals.