Inozyme Pharma and GACI Global Initiate Patient Burden of Disease Study to Improve the Understanding of the Severity and Impact of Disease in Patients with ENPP1 Deficiency and ABCC6 Deficiency

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Boston, Mass., May 14, 2020 – Inozyme Pharma, Inc., a rare disease biopharmaceutical company developing novel therapeutics for the treatment of abnormal mineralization impacting the vasculature, soft tissue and skeleton, and GACI Global (www.gaciglobal.org), a patient advocacy organization dedicated to bettering the lives of families affected by Generalized Arterial Calcification of Infancy and/or Autosomal Recessive Hypophosphatemic Rickets Type 2 (GACI/ARHR2), announced today the initiation of a study titled *Understanding the Spectrum of ENPP1 deficiency and Acute ABCC6 deficiency Through the Eyes of Patients and Parents; Burden of Illness Perspectives from Patients and Parents Who Speak English, French or German:* https://www.engagehealth.com/survey/TakeSurvey.aspx?SurveyID=8252n62#.

The purpose of the study is to characterize the burden of disease and understand the systemic progression of disease for the rare genetic diseases of both ENPP1 deficiency and ABCC6 deficiency from the perspective of a patient and/or parent. This study is designed to contribute to the overall understanding of these rare abnormal mineralization disorders and provide important clinical information regarding patient reported signs, symptoms and outcomes in order to develop appropriate endpoints in the clinical development process.

ENPP1 deficiency is a spectrum of disease which manifests as GACI type 1 in infants, ARHR2 in young adults, and Osteomalacia in older adults. ABCC6 deficiency is a disease that can lead to an acute form called GACI type 2 in infants and Pseudoxanthoma Elasticum (PXE) in older patients.

“We are excited to be initiating this important study that will characterize ENPP1 and ABCC6 deficiency from a patient and parent perspective. Inozyme is committed to the patient community and values this partnership in moving the understanding of these rare diseases forward,” said Axel Bolte, co-founder and chief executive officer of Inozyme Pharma.

“Both ENPP1 deficiency and ABCC6 deficiency are systemic and progressive disorders with high levels of morbidity and unfortunately high mortality for infants with ENPP1 deficiency,” said Pedro Huertas, M.D., Ph.D., chief medical officer of Inozyme Pharma. “Both of these deficiencies are often misdiagnosed and present as a continuum of progressive signs and symptoms throughout life. We look forward to hearing directly from patients and caregivers affected by these rare, life-threatening disorders so we can improve our understanding of the burden of these diseases. We thank GACI Global for its partnership, and we look forward to working together.”

“Participation in this patient survey study will provide a better medical understanding of the ways that ENPP1 deficiency and acute ABCC6 deficiency impacts patients and families in terms of disease burden and quality of life,” said Christine O’Brien, co-president of GACI Global. “We encourage patients and families affected by either ENPP1 deficiency or ABCC6 deficiency to learn more about this patient survey study and to participate if they are interested and able to do so. It is important that we build an understanding of both ENPP1 deficiency and ABCC6 deficiency so that these disorders are more clearly defined."

Inozyme’s lead investigational therapy, INZ-701, is an enzyme replacement therapy in development for the potential treatment of patients with ENPP1 deficiency and ABCC6 deficiency. Inozyme is preparing to seek regulatory approval to initiate a clinical trial in patients with ENPP1 deficiency and a separate clinical trial in patients with ABCC6 deficiency.

If you are interested in learning more about the study or participating in the study, click here https://www.engagehealth.com/survey/TakeSurvey.aspx?SurveyID=8252n62# or visit www.clincaltrials.gov, using the identifier NCT04372446.

About the Patient Survey Study

This patient survey is a comprehensive, prospectively designed, cross-sectional study planned for approximately 60 individuals (or representative parents of patients) affected by ENPP1 deficiency and the acute phase of ABCC6 deficiency. All study participants will participate in an interview to provide information about the burden of the disease and its impact on quality of life. The study will use validated instruments adapted for ENPP1 deficiency.

The participants will be split into four groups as described below:

- Acute infantile ENPP1 deficiency, commonly diagnosed as GACI type 1
- Acute infantile ABCC6 deficiency, commonly diagnosed as GACI type 2
- Progressive pediatric ENPP1 deficiency, commonly diagnosed as ARHR2
- Adult ENPP1 deficiency, commonly diagnosed as ARHR2

About INZ-701

INZ-701 is an enzyme replacement therapy in development for the treatment of mineralization disorders of the circulatory system, bones, and kidneys. In preclinical studies, the experimental therapy has shown potential to generate plasma pyrophosphate (PPi) and to restore it to appropriate physiological levels, thereby preventing calcification in the vasculature and kidneys, while at the same time normalizing bone mineralization. Inozyme is developing INZ-701 for certain rare, life-threatening, and devastating genetic disorders such as ENPP1 deficiency and ABCC6 deficiency in which PPi levels are below the normal physiological levels.

About Inozyme Pharma

Inozyme Pharma is a rare disease biopharmaceutical company developing novel therapeutics for the treatment of diseases of abnormal mineralization impacting the vasculature, soft tissue and skeleton. Through our in-depth understanding of the biological pathways involved in mineralization, we are pursuing the development of potentially first-in-class therapeutics to address the underlying causes of these debilitating diseases. It is well established that two genes, ENPP1 and ABCC6, play key roles in a critical mineralization pathway and that defects in these genes lead to abnormal mineralization. We are initially focused on developing a novel therapy to treat the rare genetic diseases of ENPP1 and ABCC6 deficiencies.

Inozyme Pharma was founded in 2017 by Joseph Schlessinger, Ph.D., Demetrios Braddock, M.D., Ph.D., and Axel Bolte, MSc, MBA, with technology developed by Dr. Braddock and licensed from Yale University. For more information, please visit www.inozyme.com.

About GACI Global

GACI Global is a nonprofit patient advocacy group whose mission is to connect families affected by Generalized Arterial Calcification of Infancy of Hypophosphatemic Rickets caused by ENPP1 or ABCC6 deficiencies to each other and to the medical community. The organization strives to provide current educational resources and supports ongoing research. The goal of this 100% volunteer run organization is to provide not only information about what complications can occur due to ENPP1 and ABCC6 deficiencies, but to provide hope for families impacted by the condition around the world.

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