Inozyme Pharma Announces Peer-Reviewed Publication Revealing Increased Prevalence of ENPP1 Deficiency

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- Article in Orphanet Journal of Rare Diseases *estimates genetic prevalence of ENPP1 Deficiency at 1 in 64,000 pregnancies, more than tripling prior estimate -*

- Company estimates addressable patient population with ENPP1 Deficiency at 37,000 worldwide; 2,800 in North America, 4,100 in Europe and 900 in Japan -

BOSTON, Dec. 05, 2022 (GLOBE NEWSWIRE) -- Inozyme Pharma, Inc. (Nasdaq: INZY), a clinical-stage rare disease biopharmaceutical company developing novel therapeutics for the treatment of pathologic mineralization and intimal proliferation, today announced the publication of an article titled "Estimation of ENPP1 Deficiency Genetic Prevalence Using a Comprehensive Literature Review and Population Databases" in the *Orphanet Journal of Rare Diseases.* Leading disease experts Carlos Ferreira, M.D., of the National Institutes of Health (NIH) and Frank Rutsch, M.D., of Münster University Children's Hospital, together with Genomenon, an AI-driven genomics company, analyzed the latest genomics data and found the estimated prevalence of ENPP1 Deficiency to be 1 in 64,000 pregnancies, more than tripling the prior estimate[1].

"Our medical field team's disease awareness, education, and patient identification efforts suggested that the number of patients with ENPP1 Deficiency has been vastly underestimated. These new data provide an independent and thorough calculation supporting our observations and highlight the urgent need for a therapeutic option," said Axel Bolte, MSc, MBA, Inozyme's co-founder, president and chief executive officer.

The genetic prevalence was calculated using American College of Medical Genetics guidelines to interpret the latest data on disease-causing *ENPP1* variants and to calculate carrier frequency with the Genome Aggregation Database (gnomAD).

"Given the higher prevalence of ENPP1 Deficiency, it becomes obvious that many affected individuals remain undiagnosed. These new data should alert physicians and increase the awareness and diagnosis of GACI and ARHR2," added Frank Rutsch, M.D., professor of pediatrics at Münster University Children's Hospital.

"This latest dataset further validates the previously published patient and variant database for ENPP1 Deficiency," said Mark Kiel, M.D., Ph.D., chief science officer and co-founder of Genomenon. "The prevalence data were gathered using a comprehensive database of genomic evidence, providing an unprecedented view into how many people are born with ENPP1 Deficiency worldwide. We expect that discovery of this much higher prevalence rate, reinforced by continuous updates to the database, will facilitate more accurate and timely diagnoses."

About ENPP1 Deficiency

ENPP1 Deficiency is a progressive condition that manifests as a spectrum of diseases. Individuals who present in utero or in infancy are typically diagnosed with generalized arterial calcification of infancy (GACI), which is characterized by extensive vascular calcification and neointimal proliferation (overgrowth of smooth muscle cells inside blood vessels), resulting in myocardial infarction, stroke, or cardiac or multiorgan failure. Approximately 50% of infants with ENPP1 Deficiency die within six months of birth. Children with ENPP1 Deficiency typically experience rickets, a condition also known as autosomal-recessive hypophosphatemic rickets type 2 (ARHR2), while adults experience osteomalacia (softened bones), and they can exhibit a range of signs and symptoms that include hearing loss, arterial calcification, and cardiac and/or neurological involvement. There are no approved therapies for ENPP1 Deficiency**.**

About Inozyme Pharma

Inozyme Pharma, Inc. (Nasdaq: INZY) is a clinical-stage rare disease biopharmaceutical company developing novel therapeutics for the treatment of diseases impacting the vasculature, soft tissue, and skeleton. We are developing INZ-701, a potential first-in-class enzyme therapy, to address pathologic mineralization and intimal proliferation which can drive morbidity and mortality in these severe diseases. INZ-701 is currently in Phase 1/2 clinical trials for the treatment of ENPP1 Deficiency and ABCC6 Deficiency.

For more information, please visit www.inozyme.com and follow us on LinkedIn, Twitter, and Facebook.

About Genomenon

Genomenon is an AI-driven genomics company focused on making genomic information actionable for patients with rare genetic diseases and cancer. Keeping pace with the ever-evolving body of knowledge within genomics, Genomenon connects current research with patient DNA to accelerate clinical decision-making and pharmaceutical drug discovery. For more information, visit Genomenon.com

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[1] Ferreira et al. Genet. Med, 2020.