Inozyme Pharma Announces Publication of Comprehensive ENPP1 Variant Database

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- Peer-reviewed article in Human Mutation *reports 3-fold increase in pathogenic/likely pathogenic ENPP1 variants –*

- Database identified severely symptomatic patients with monoallelic heterozygous ENPP1 variants –

BOSTON, Oct. 31, 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, "ENPP1 Deficiency: A clinical update on the relevance of individual variants using a locus-specific database," in *Human Mutation.* The Company partnered with leading disease experts and Genomenon, an AI-driven genomics company, to perform a comprehensive review of the current knowledge on clinical and genetic findings of ENPP1 Deficiency (GACI, generalized arterial calcification of infancy or ARHR2, autosomal recessive hypophosphatemic rickets type 2) and produce a comprehensive variant database.

"Our team continues to apply innovative methods to better understand ENPP1 Deficiency to increase disease awareness and inform our clinical and regulatory efforts, as we work to develop the first potential treatment for this rare and devastating lifelong disease," said Henric Bjarke, Inozyme's senior vice president and chief operating officer. "These data shared by our collaborators suggest a larger population with ENPP1 Deficiency than previously understood. The publication also showed that there was no clear genotype-phenotype correlation and that people with heterozygous mutations may also benefit from a therapeutic option."

The review analyzed all published cases of ENPP1 Deficiency (n=154) and results from two natural history studies of GACI and ARHR2 patients.[1] The associated genetic variants were interpreted using Genomenon's Mastermind ® Genomic Search, a database of variants with evidence cited in medical literature. The database is used by more than 2,000 genetic testing laboratories and medical centers across the globe and connects patient DNA to relevant scientific research to enable diagnosis and treatment decisions.

Summary of Key Results

- 109 unique *ENPP1* variants discovered with 79 identified as pathogenic/likely pathogenic, representing a 3-fold increase in the number of pathogenic/likely pathogenic variants compared to other databases.
- Data suggested no genotype-phenotype correlation.
- Analysis identified severe phenotypes in patients with monoallelic heterozygous *ENPP1* variants.

"Genomenon has worked with Inozyme for the past year to produce a comprehensive variant landscape for ENPP1 Deficiency," said Mark Kiel, M.D., Ph.D., chief science officer and co-founder of Genomenon. "The AI-driven genetic dataset, along with information on available clinical trials, can be accessed by doctors, researchers, and clinicians through Genomenon's Mastermind ® Genomic Search Engine, and has the potential to increase the number of patients who are accurately diagnosed with this devastating rare disease. Our partnership with Inozyme puts critical information about ENPP1 Deficiency at the fingertips of doctors via a database that is continually updated as new evidence is published and will play a key role in addressing missed diagnoses, which has long been a challenge for this disease community."

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 first-in-class enzyme therapy, to address pathologic mineralization and intimal proliferation which 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 the advancement of positive health outcomes 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.

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[1] Ferreira, Kintzinger, et al., (2021). Ectopic calcification and hypophosphatemic rickets: natural history of ENPP1 and ABCC6 deficiencies. *Journal of Bone and Mineral Research*, **36**(11), 2193–2202. https://doi.org/10.1002/jbmr.4418