



Inozyme Pharma Offers No-Cost Genetic Testing for Rare Calcification Disorders

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CAMBRIDGE, Mass., June 13, 2018 – [Inozyme Pharma](#) (Inozyme), a biopharmaceutical company dedicated to developing treatments for rare and debilitating diseases, today introduced a no-cost, third-party genetic testing program designed to improve detection and understanding of two rare calcification disorders. Offered globally and in partnership with [PreventionGenetics](#), the Inozyme program tests eligible participants for mutations in the *ENPP1* and *ABCC6* genes.

Both of these genes are implicated in rare, severe calcification disorders, known as *ENPP1* deficiency and *ABCC6* deficiency, respectively, (sometimes called generalized arterial calcification of infancy [GACI] and autosomal recessive hypophosphatemic rickets type 2 [ARHR2]).

“The genetic testing program introduced today will help to enhance our understanding of *ENPP1* deficiency and *ABCC6* deficiency, with the ultimate goal of improving diagnosis and developing effective treatments,” said Axel Bolte, co-founder and chief executive officer of Inozyme. “We urge people with a family history of calcification disorders and their physicians to seek more information about the genetic testing program. Over time and through ongoing research, we hope to alleviate the life-limiting and life-threatening impact of these severe diseases.”

Inozyme created the genetic testing program to increase disease awareness, reduce barriers to genetic testing, and help people and their healthcare providers make more informed decisions about these rare conditions.

“The Inozyme genetic testing program will provide an early diagnostic testing measure for *ENPP1* and *ABCC6* deficiencies, which are indicators of serious calcification disorders that sometimes take years to diagnose accurately,” said James Weber, president of PreventionGenetics. “Access to reliable genetic testing may help shorten the diagnostic journey for patients, potentially lifting a significant emotional burden and paving the way for more timely and effective intervention. We are excited to work with Inozyme as we make the test available to physicians and their patients.”

The no-cost program offered by Inozyme and PreventionGenetics provides genetic testing to those who qualify. Although genetic testing can confirm a suspected diagnosis of a calcification disorder linked to *ENPP1* or *ABCC6* gene mutations, the absence of a genetic alteration does not preclude diagnosis of such a disease. For more information about the genetic testing program, please visit: www.inozyme.com/genetic-testing/.

About *ENPP1* Deficiency

The *ENPP1* gene produces a critical enzyme called ectonucleotide pyrophosphatase/ phosphodiesterase 1 (*ENPP1*), which regulates inorganic pyrophosphate (PPi) levels in plasma. PPi is essential for preventing harmful soft tissue calcification and for regulating normal bone mineralization. *ENPP1* deficiency manifests as either generalized arterial calcification of infancy (GACI) type 1 or autosomal recessive hypophosphatemic rickets type 2 (ARHR2). GACI type 1 is a devastating and often fatal disease affecting infants and is characterized by calcification and narrowing of large and medium-sized arteries, resulting in heart failure and death in about half of patients within the first six months of life. ARHR2 usually manifests in the post-infancy stage, though it can occur in patients without prior GACI. ARHR2 causes rickets, weakened bones, repeated bone fractures, skeletal deformities, short stature, muscle weakness, fatigue and bone pain.

About *ABCC6* Deficiency

Defects in the *ABCC6* gene (ATP-binding cassette sub-family C member 6) lead to a decrease in plasma PPi and consequently to soft tissue calcification, and in rare circumstances cause GACI type 2 in infants. GACI type 2 is clinically similar to GACI type 1 and is also characterized by calcification and narrowing of large and medium-sized arteries, resulting in heart failure and death in about half of patients within the first six months of life.

About Inozyme Pharma

Inozyme Pharma is a biotechnology company committed to developing novel medicines for the treatment of rare diseases characterized by mineral imbalances, which lead to over-calcification of soft tissues and under-mineralization of bone. The company was founded in 2016 with technology licensed from Yale University. For more information, please visit: www.inozyme.com.

About PreventionGenetics

PreventionGenetics is a CLIA and ISO 15189:2012 accredited clinical DNA testing laboratory founded in 2004 and located in Marshfield, Wisconsin. PreventionGenetics provides patients with sequencing and deletion/duplication tests for nearly all clinically relevant genes, including whole exome sequencing, PGxome®.

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