Inozyme's No-Cost Genetic Testing Program for ENPP1 and ABCC6 Deficiencies

In partnership with PreventionGenetics, Inozyme offers a global, no-cost genetic testing program for the *ENPP1* and *ABCC6* genes; both of these genes are implicated in two 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 Process

You will determine if the individual meets eligibility criteria and discuss the test.

You will order the test and collect a blood or saliva sample and ship it to the lab.

The genetic test will be processed at PreventionGenetics and the results will be sent to you about 14 days after the lab receives the sample and all appropriately completed paperwork. You will discuss the results with the patient and/or caregiver.

Eligibility Criteria and Ordering the Test

If you have a patient you suspect to have one of these conditions or to be a carrier, they may be eligible for the no-cost genetic test.

You can obtain information about eligibility criteria and ordering the genetic test using the below information:

1-715-387-0484, choose option 2 and mention Program #SP-051

https://www.preventiongenetics.com/specialOrders?sp=sp051

Insurance and Privacy Information

Will the test require a payment from my patient or their insurance company?

No, neither your patient nor their insurance will pay for this test. Inozyme Pharma is sponsoring the genetic testing program at no charge to your patient or their insurance company. Inozyme will pay for the test whether a mutation is found in either gene or not.

Will patient's results be kept confidential?

Your patient's test results will be kept confidential. Your patient's genetic test results will not be shared with their insurance company, Inozyme or others. Inozyme will receive information regarding the overall test results for the program but will not be given any identifying information such as your patient's name, date of birth, or address. Inozyme will receive your name and contact information to contact you with information about Inozyme's clinical development in ENPP1 Deficiency and ABCC6 Deficiency programs such as natural history studies, patient registries, and clinical trials. Inozyme's Privacy Policy can be found at http://www.inozyme.com/privacy-policy/.

ENPP1 Deficiency: GACI Type 1 and/or ARHR2

GACI Type 1 and 2 and AHRH2 are inherited in an autosomal recessive manner, meaning that an individual needs to inherit two mutations, one from their mother and one from their father, to have ENPP1 or ABCC6 Deficiency.

GACI Type 1 is a rare, devastating and potentially fatal disease, characterized by extensive calcifications and narrowing of the large and medium arteries, resulting in heart failure and death in about half of the patients within the first six months of life.

ARHR2 manifests post-infancy and is characterized by typical clinical features of rickets such as weakened bones, skeletal deformities, short stature, and bone pain. During adulthood, ARHR2 clinical findings may include bone pain, fatigue, muscle weakness, and repeated bone fractures.

ABCC6 Deficiency: GACI Type 2

GACI Type 2 is clinically similar to GACI type 1, 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.

Inozyme Pharma is a biotechnology company committed to developing novel medicines for the treatment of rare metabolic diseases of calcification. The company was founded in 2016 with technology licensed from Yale University. For more information, please visit: www.inozyme.com

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