



Hypoparathyroidism

HYPOPARATHYROIDISM GENETIC TESTING PROGRAM DESCRIPTION

Invitae and Calcilytix Therapeutics Inc. have partnered to offer sponsored, no-charge genetic testing for individuals with an established diagnosis of non-surgical hypoparathyroidism. An accurate diagnosis of genetic hypoparathyroidism may have an impact on clinical management of the condition.

This **Hypoparathyroidism Genetic Testing program** was created to learn more about the genetic causes of hypoparathyroidism and support clinical management so that individuals and their providers can make more informed decisions about their health.

For more information or to order a test, please visit
www.invitae.com/hypoparathyroidism

ABOUT HYPOPARATHYROIDISM



Hypoparathyroidism is estimated to affect approximately 37 in every 100K individuals in the US.¹



Symptoms of hypoparathyroidism can include hypocalcemia-related neuromuscular (i.e. cramps, paresthesia, and/or seizures), cardiovascular, and neurological (i.e. fatigue and/or cognitive impairment) manifestations.¹



An accurate diagnosis of genetic hypoparathyroidism may have an impact on a patient's clinical management.

PANEL INFORMATION

This program offers the **Invitae Hypoparathyroidism and Hyperparathyroidism Panel**, which tests for variants in 7 genes known to be associated with various hereditary causes of parathyroid disorder.

**This test is specifically designed for heritable germline mutations and is not appropriate for the detection of somatic mutations in tumor tissue.*

STANDARD CLINICAL CONSULT SERVICES AVAILABLE

Invitae's team of board-certified and experienced genetic counselors trained in medical genetics is available to assist clinicians and patients by phone and/or email. Our genetic counselors can be reached throughout the testing process to:

- review patient cases that may benefit from the Program
- aid in interpreting results
- provide result-specific background regarding variants, genes, and the condition
- identify gene-specific information including relevant literature and studies, published management guidelines if available, and patient resources

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While third parties, including Calcilytix Therapeutics Inc., and commercial organizations may provide financial support for this program, tests and services are performed by Invitae. Healthcare professionals must confirm that patients meet certain criteria to use the program. Third parties, including Calcilytix Therapeutics Inc., and commercial organizations may receive de-identified patient data from this program, but at no time would they receive patient identifiable information. Third parties, including Calcilytix Therapeutics Inc., and commercial organizations may receive contact information for healthcare professionals who use this program. Genetic testing is available in the US only. Healthcare professionals and patients who participate in this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any other products or services from Invitae or from third parties, including Calcilytix Therapeutics Inc., or commercial organizations.

Reference:

1. LSinnott BP (2018), Hypoparathyroidism – Review of the Literature 2018. J Rare Disord Diagn Ther. Vol.4 No.3:12