Uncovering Periodic Paralysis

Genetic testing to shorten the diagnostic journey

Talk to your healthcare provider to request a genetic test for Periodic Paralysis.

For more information, please visit:

www.UncoveringPeriodicParalysis.com

No-Cost Periodic Paralysis Genetic Testing program

Strongbridge Biopharma® has partnered with Invitae to offer a Periodic Paralysis genetic test for those who have episodes of muscle weakness or temporary paralysis provoked by common triggers for primary hyperkalemic or hypokalemic periodic paralysis.

A Periodic Paralysis genetic test can bring you closer to uncovering the genetic cause behind the attacks of paralysis and muscle weakness caused by hyperkalemic or hypokalemic periodic paralysis.

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Periodic Paralysis Genetic Test

- The Invitae Periodic Paralysis genetic test includes analysis for both hyperkalemic and hypokalemic periodic paralysis.
    - Testing with the Invitae Periodic Paralysis genetic test includes analysis of the following six most common associated genes: **ATP1A2, SCN4A, CACNA1S, KCNJ2, MCM3AP, and RYR1**
- Turnaround time for the test is rapid: Once Invitae receives the sample, your doctor will receive results within 10 to 21 calendar days (14 days on average).
- Invitae's board-certified genetic counselors are available on-demand Monday through Friday, 5 am to 5 pm Pacific Time.
- To have a genetic test completed, you must provide a sample of your saliva or blood.

Program Eligibility

- 18 years of age and older.
- Episodes of muscle weakness and/or paralysis attacks or episodes of pain after attacks (happens at least more than once).
- Episodes are caused by at least one of the common triggers for hyperkalemic or hypokalemic primary periodic paralysis.

Utilizing the *Uncovering Periodic Paralysis* program imposes no obligation to prescribe or use Strongbridge Biopharma products or services.

How To Get a Genetic Test

Your healthcare provider must order the test for you. Once ordered, you will need to provide a sample of your saliva or blood for testing.

How to provide your sample:

1. Your healthcare provider can give you a kit to collect either your saliva OR blood
2. Ability to schedule a no-cost blood draw appointment at a doctor's office or at your home (within the U.S.)

Family Variant Testing

- Strongbridge Biopharma and Invitae offer no-cost family variant testing to any blood relative of anyone newly diagnosed with PPP through the *Uncovering Periodic Paralysis* genetic testing program.
- Requirements:
  - The *Uncovering Periodic Paralysis* genetic test results must show a positive diagnosis with one of the six known PPP genes (ATP1A2, SCN4A, CACNA1S, KCNJ2, MCM3AP, RYR1)
  - A doctor must place the order within 90 days of the original test report

About Primary Periodic Paralysis (PPP)

- Primary Periodic Paralysis is very rare, affecting ~4,000 to 5,000 diagnosed individuals in the US.[1]
- This condition causes attacks that affect your muscles, resulting in extreme weakness or temporary paralysis—most often the inability to move the muscles of the arms and legs.[2]
- Primary Periodic Paralysis is usually inherited from a parent and may affect multiple people within a family.[3]

A negative test does not rule out a PPP diagnosis[4]

A negative test result may still mean that you have PPP. Genetic testing can confirm a diagnosis in about 70% of those who have PPP, but the remaining 30% don't have one of the commonly identified genetic mutations, but are still considered to have PPP based on symptoms, family, history, and/or other tests.

Healthcare providers should also consider the below areas when making the diagnosis[4]:

- Clinical presentation of symptoms
- History of attacks (as well as serum potassium levels during attacks)
- Response to specific PPP triggers
- Family history
- Ruling out other conditions that mimic PPP
- Other testing such as ECG/EKG, EMG, and CMAP

Interested in learning more?
Visit: www.PPPDiagnosis.com
to read an *Expert Roundtable on Primary Periodic Paralysis*

References:
1. Data on File. Available from Strongbridge Biopharma plc. **2.** Charles G, Zheng C, Lehmann-Horn F, Jurkat-Rott K, Levitt J. Characterization of hyperkalemic periodic paralysis: a survey of genetically diagnosed individuals. *J Neurol.* 2013;260:2606-2613. **3.** Fontaine B, Phillips LH 2nd. A newly approved drug for a rare group of diseases: dichlorphenamide for periodic paralysis. Neurology. 2016;86:1366-1367. **4.** Statland JM, Fontaine B, Hanna MG, et al. Review of the diagnosis and treatment of periodic paralysis. Muscle Nerve. 2018;57:522-530

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Strongbridge CareConnection offers a wide range of services for patients and their families

One-on-One Support

- A dedicated case manager is assigned to every patient
- Patient access managers are available to work directly with patients and healthcare providers to overcome common barriers to therapy
- Patients receive routine calls from a licensed pharmacist, which focus on progress and goals of therapy and disease management

Treatment Access Assistance

- Support in understanding insurance coverage
- Information on a $0 co-pay program and other financial assistance
- Access to a patient assistance program for patients without insurance coverage
- Convenient, free specialty pharmacy services
    - Home delivery of prescriptions and refills
    - 24/7 support

Patient Education

- Information about Primary Periodic Paralysis
- Information about a treatment option

Community Connection

- Links to events sponsored by advocacy organizations
- Online patient communities

Connect with the CareConnection Team by calling 844-538-3947

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