Uncovering Periodic Paralysis

A sponsored no-cost Primary Periodic Paralysis (PPP) genetic testing program

PPP is a rare and potentially progressive genetic condition.[1-3] Strongbridge Biopharma® has partnered with Invitae to offer a periodic paralysis genetic test for those who have episodic muscle weakness or temporary paralysis provoked by common triggers for PPP.

A genetic test can help shorten the diagnostic journey and expedite clinical management of the disease.

ORDER A TEST TODAY

www.uncoveringperiodicparalysis.com

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PPP is a rare inherited condition[3]

- PPP includes several autosomal dominant inherited neuromuscular disorders which cause **recurrent, progressive, and debilitating episodes of extreme muscle weakness and temporary paralysis**[3-4]
  - Although there are other related variants, the most common forms of PPP are hypokalemic periodic paralysis and hyperkalemic periodic paralysis[5-8]

PPP IS OFTEN DIFFICULT TO DIAGNOSE[1-2]

- Patients in one study reported seeing an average of 4 doctors before receiving a correct diagnosis of PPP[1]*
- Average time from onset of symptoms to correct diagnosis **26 years**†[2]

*Based on a survey of 137 patients ages 19-84 years with a diagnosis of hyperkalemic periodic paralysis who were invited through the Internet or one of several healthcare institutions worldwide.

† Based on a survey of 66 self-selected patients over the age of 40 years with a clinical diagnosis of Primary Periodic Paralysis who sought support via the Internet.

PPP IS COMMONLY MISDIAGNOSED[1,2,9]

- Patients have reported being misdiagnosed with other conditions, including depression, malingering, conversion disorder, or myotonia congenita[1]

A negative test does not rule out a PPP diagnosis[3]

Patients with a negative test result may still have PPP. Genetic testing can confirm a diagnosis in about 70% of patients, but the remaining 30% don't have one of the commonly identified genetic mutations.

These patients can be diagnosed by considering[3]:

- 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 A NEW DIAGNOSTIC ALGORITHM

STRONGBRIDGE™ CareConnection

Provides a range of support for patients

Genetic testing is a simple, convenient process

1. **Place your order:**
Online Order: Place an order through the ordering portal at **www.uncoveringperiodicparalysis.com**
-or-
Fax Order: Fill out the enclosed form, which is also available at **www.uncoveringperiodicparalysis.com,** and fax it to 415-276-4164

2. **Collect a specimen:** Order a specimen collection kit at **www.invitae.com/request-a-kit**. Make sure to label the sample with the patient's full name, date of birth, and sample collection date. Your Strongbridge Area Business Specialist can also provide you with a kit.

3. **Get the results:** Once Invitae receives the sample, you will receive the results in 10-21 calendar days, on average. If you create an online account, you will be able to track the status of your order and receive a notification email when the test results are ready.

Eligibility for Testing?

- Patients must be 18+ years of age
- Have a history of episodic muscle weakness, paralysis attacks, or episodic pain after attacks (more than 1 occurrence)
- Have episodes provoked by at least 1 of the common triggers for PPP. Visit www.uncoveringperiodicparalysis.com to view the common triggers of PPP.

No-Cost Testing For Family Members*

- Strongbridge Biopharma and Invitae offer no-cost family variant testing to any blood relative of a patient newly diagnosed through the Uncovering Periodic Paralysis genetic testing program
- Eligibility for family members:
  - A pathogenic or likely pathogenic variant was found on the original patient's Uncovering Periodic Paralysis gene panel test
  - The order for the genetic test must be placed within 90 days of the original patient's test report

–*Direct family members do **not** have to meet any of the eligibility requirements (age, triggers, episodes, etc.) to receive family variant testing.

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The advantages of the PPP genetic testing panel

- **Screens for hyperkalemic, hypokalemic, and atypical periodic paralyses, as well as Andersen-Tawil syndrome,** including the 6 most commonly associated genes: *ATP1A2, SCN4A, CACNA1S, KCNJ2, MCM3AP, and RYR1.*
- On the requisition form, you have the opportunity to check a box that will automatically reflex to analysis using the Invitae Comprehensive Neuromuscular Disorders Panel should the initial result be negative. However, if you don't check that box, you still have 90 days to order re-requisition analysis.
- **Turnaround time for the panel is rapid.** Once Invitae receives the sample, you will receive the results in 10-21 calendar days, on average.
- **Invitae's board-certified genetic counselors are available** Monday through Friday, 5 AM to 5 PM Pacific time, to help review patient cases, differentiate between test options, and aid in interpreting results. **If you have questions, you can reach the Invitae Customer Service team at 800-436-3037**

A genetic test can help shorten the PPP diagnostic journey and expedite clinical management of the disease

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900 Northbrook Drive, Suite 200
Trevose, PA 19053 United States
www.strongbridgebio.com

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