A no-cost Primary Periodic Paralysis (PPP) gene panel testing program

PPP is a rare and potentially progressive genetic condition.1-5 Strongbridge Biopharma® has partnered with Invitae to offer a periodic paralysis gene panel 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
PPP includes several autosomal dominant inherited neuromuscular disorders that are usually chronic. These disorders cause recurrent, progressive, and debilitating episodes of extreme muscle weakness and temporary paralysis\(^1,2,6\)

- Although there are other related variants, the most common forms of PPP are hypokalemic periodic paralysis and hyperkalemic periodic paralysis\(^7,11\)

- PPP may affect multiple individuals within a family\(^12\)

**It can be a long and frustrating journey to reach a PPP diagnosis**

**PPP IS OFTEN DIFFICULT TO DIAGNOSE\(^4\)**

- Patients in one study reported seeing an average of 4 doctors before receiving a correct diagnosis of PPP*

- For some patients, diagnosis has taken up to 20 years

*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.

**PPP IS COMMONLY MISDIAGNOSED\(^1,4\)**

- Patients have reported being misdiagnosed with other conditions, including cardiovascular disorders, or mental disorders such as depression, before receiving their diagnosis

**A negative test does not rule out a PPP diagnosis\(^1\)**

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\(^1\):

- 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
**Genetic testing is a simple, convenient process**

1. **Place your order:** Fill out the enclosed form, which is also available at [www.uncoveringperiodicparalysis.com](http://www.uncoveringperiodicparalysis.com), and fax it to 415-276-4164.
   - Place an order through the ordering portal at [www.uncoveringperiodicparalysis.com](http://www.uncoveringperiodicparalysis.com)

2. **Collect a specimen:** Order a specimen collection kit at [www.invitae.com/request-a-kit](http://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.

3. **Get the results:** You will have results in hand 10 to 21 days after Invitae receives the sample. 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.

**Who is eligible?**

**ADULTS:**
- 18+ years of age
- With episodic muscle weakness/paralysis attacks or episodic pain after attacks (more than 1 occurrence)
- Who have episodes provoked by at least 1 of the common triggers for PPP

**Direct family members can get tested, too**
- 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
- **Requirements:**
  - A pathogenic or likely pathogenic variant was found on the original patient’s Uncovering Periodic Paralysis gene panel test
  - The order must be placed within 90 days of the original patient’s test report
The advantages of the PPP genetic testing panel

- Screens for hyperkalemic, hypokalemic, and atypical periodic paralyses, as well as Andersen-Tawil syndrome, including the 4 most commonly associated genes: SCN4A, CACNA1S, KCNJ2, and RYR1

- If medically appropriate, re-requisition to the larger Invitae Comprehensive Neuromuscular Disorders panel within 90 days of original test report

- **Turnaround time for the panel is rapid.** Once Invitae receives the sample, you will receive results within 10 to 21 calendar days (14 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

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ORDER A KIT TODAY

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