Uncovering Periodic Paralysis

Genetic testing to shorten the diagnostic journey

For more information, please visit:
www.UncoveringPeriodicParalysis.com

No-cost Periodic Paralysis gene panel testing program

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 primary hyperkalemic or hypokalemic periodic paralysis.

A Periodic Paralysis gene panel test can bring you and your patients closer to uncovering the genetic cause behind the attacks of paralysis and muscle weakness caused by hyperkalemic or hypokalemic periodic paralysis. It can help shorten the diagnostic journey and expedite clinical management of the disease.

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Periodic Paralysis Panel

- The Invitae Periodic Paralysis Panel includes analysis for both hyperkalemic and hypokalemic periodic paralysis.

  Testing with the Invitae Periodic Paralysis Panel includes analysis of the following three most common associated genes: **SCN4A, CACNA1S, and KCNJ2**

- 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 on-demand Monday through Friday, 5 am to 5 pm Pacific Time, to help review patient cases and aid in interpreting results.

Family Variant Testing

- Strongbridge Biopharma and Invitae offer no-cost family variant testing to any first-degree 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 Uncovering Periodic Paralysis gene panel test
  - Order must be placed within 90 days of the original test report

Program Eligibility

- **18+ years of age**
- **Episodic muscle weakness/paralysis attacks or episodic pain after attacks (more than one occurrence)**
- **Episodes are provoked by at least one of the common triggers for hyperkalemic or hypokalemic primary periodic paralysis**

Hyperkalemic Primary Periodic Paralysis

Associated with paralytic episodes with concomitant hyperkalemia (serum potassium concentration >5 mmol/L) or a serum potassium increase of at least 1.5 mmol/L during an attack[1]

Triggers may include

- Cold environment
- Rest after exercise
- Stress or fatigue
- Alcohol and certain foods/beverages
- Potassium-rich food
- Hunger/fasting
- Changes in activity level
- Changes in humidity
- Extra sleep
- Pregnancy and menstruation
- Illness of any type
- Some medications
- Potassium supplements

Hypokalemic Primary Periodic Paralysis

Associated with paralytic episodes with concomitant hypokalemia (<2.5 mmol/L)[2]

Triggers may include

- Rest after exercise
- Carbohydrate-rich evening meals
- Periods of inactivity, including nocturnal rest
- Heavy meals
- Sweets
- Salt
- Cold
- Stress

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

Ordering Made Easy

Place your order

To place a paper-based order, fill out the enclosed requisition form or download the form found at: **www.UncoveringPeriodicParalysis.com** and fax to 415-276-4164

To place an order online, enter the online ordering portal found at www.UncoveringPeriodicParalysis.com

Collect a specimen

To collect a specimen:

1. Order a specimen collection kit at www.invitae.com/request-a-kit
2. Label the sample tube with the patient's full name, date of birth, and sample collection date
3. For additional information see specimen and shipping requirements at www.invitae.com/specimen-requirements

Results

To receive results:

4. Once Invitae receives the sample, you will receive the results in 10-21 calendar days, 14 days on average
5. If you created an online account, you can view the status of your order by logging into your account
6. You will receive a notification email once the test results are ready

Although genetic testing can help confirm a suspected diagnosis, the absence of a genetic alteration does not preclude diagnosis.[1,2]

Convenient Specimen Collection

Resources Offered:

1. No-cost kits for blood or saliva specimen collection.
2. Phlebotomy services at no additional cost within the US
3. Ability to schedule a blood draw appointment at a physician's office or at a patient's home

About Primary Periodic Paralysis (PPP)

- Primary Periodic Paralysis is very rare, affecting ~4,000 to 5,000 diagnosed individuals in the US.[3]
- This condition includes a spectrum of rare and chronic genetic, neuromuscular disorders with autosomal dominant inheritance that cause recurrent, progressive, and debilitating episodes of extreme muscle weakness and temporary paralysis that may negatively impact patients' daily lives.[4-8]
- Although there are related variants, the most common forms are hyperkalemic and hypokalemic periodic paralysis.[9-13]
- Primary Periodic Paralysis is usually inherited from a parent and may affect multiple individuals within a family.[14]

References:

1. Weber F, Jurkat-Rott, Karin, Lehmann-Horn F. Hyperkalemic Periodic Paralysis. NCBI Bookshelf. A service of the National Library of Medicine, National Institutes of Health. 2016. **2.** Vicart S, Sternberg D, Arzel-Hezode, M, et al. Hypokalemic Periodic Paralysis. NCBI Bookshelf. A service of the National Library of Medicine, National Institutes of Health. 2014. **3.** Data on File. Available from Strongbridge Biopharma plc. **4.** Greig SL. Dichlorphenamide: a review in primary periodic paralyses. *Drugs.* 2016;76:501-507. **5.** 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. **6.** Arya SN. Periodic paralysis. *Journal, Indian Academy of Clinical Medicine.* 2002;3:374-382. **7.** Cannon SC. Channelopathies of skeletal muscle excitability. *Compr Physiol.* 2015;5:761-790. **8.** Cavel-Greant D, Lehmann-Horn F, Jurkat-Rott K. The impact of permanent muscle weakness on quality of life in periodic paralysis: a survey of 66 patients. *Acta Myol.* 2012;31:126-133. **9.** National Institutes of Health. Hyperkalemic periodic paralysis. Available at: https://ghr.nlm.nih.gov/condition/hyperkalemic-periodic-paralysis. Accessed December 5, 2016. **10.** National Institutes of Health. Hypokalemic periodic paralysis. Available at: https://ghr.nlm.nih.gov/condition/hypokalemic-periodic-paralysis. Accessed December 5, 2016. **11.** National Institutes of Health. Anderson-Tawil syndrome. Available at: https://ghr.nlm.nih.gov/condition/andersen-tawil-syndrome. Accessed December 5, 2016. **12.** National Institutes of Health. Potassium-aggravated myotonia. Available at: https://ghr.nlm.nih.gov/condition/potassium-aggravated-myotonia. Accessed December 5, 2016. **13.** National Institutes of Health. Paramyotonia congenita. Available at: https://ghr.nlm.nih.gov/condition/paramyotonia-congenita. Accessed December 5, 2016. **14.** Fontaine B, Phillips LH 2nd. A newly approved drug for a rare group of diseases: dichlorphenamide for periodic paralysis. Neurology. 2016;86:1366-1367.

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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
- Treatment support from highly trained specialized healthcare professionals

Treatment Access Assistance

- Support in understanding insurance coverage
- Information on co-pay and financial assistance programs
- Access to a patient assistance program
- 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

Genetic Testing

- *Uncovering Periodic Paralysis*: No-cost Periodic Paralysis gene panel testing program
- Visit www.UncoveringPeriodicParalysis.com for more information

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