



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 Xeris Pharmaceuticals® has partnered with PreventionGenetics 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

uncoveringperiodicparalysis.com

PPP is a rare inherited condition³

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^{3,4}

PPP IS OFTEN DIFFICULT TO DIAGNOSE^{1,2} AND COMMONLY MISDIAGNOSED^{1,2,9}

- Patients in one study reported seeing an average of 4 doctors before receiving a correct diagnosis of PPP¹
- Average time from onset of symptoms to correct diagnosis is **26 years**¹²
- Patients have reported being misdiagnosed with other conditions, including depression, malingering, conversion disorder, or myotonia congenita¹

A NEGATIVE TEST DOES NOT RULE OUT A PPP DIAGNOSIS⁵

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:

- 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

^{*}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.

ECG/EKG = electrocardiogram; EMG = electromyography; CMAP = compound muscle action potential

Genetic testing is a simple, convenient process

PLACE YOUR ORDER

- 1 ORDER ONLINE** Place an order for a specimen collection kit through the ordering portal at uncoveringperiodicparalysis.com. Your Xeris Area Business Specialist can also provide you with a kit.
- 2 COLLECT A SPECIMEN** Make sure to label the sample with the patient's full name, date of birth, and sample collection date.
- 3 GET THE RESULTS** Once PreventionGenetics receives the sample, you will receive the results in 18-24 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

- 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 uncoveringperiodicparalysis.com to view the common triggers of PPP

NO-COST TESTING FOR FAMILY MEMBERS

- Xeris Pharmaceuticals® and PreventionGenetics 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
- Direct family members do **not** have to meet any of the eligibility requirements (age, triggers, episodes, etc.) to receive family variant testing

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: *ATPIA2*, *SCN4A*, *CACNA1S*, *KCNJ2*, *MCM2AP*, and *RYR1*
- Turnaround time for the panel is rapid. Once PreventionGenetics receives the sample, you will receive the results in 18-24 days, on average
- Board-certified genetic counselors are available to help review patient cases, differentiate between test options, and aid in interpreting results
- If you have questions, you can email support@preventiongenetics.com

ORDER A TEST TODAY

A genetic test can help shorten the PPP diagnostic journey and expedite clinical management of the disease
[uncoveringperiodicparalysis.com](https://www.preventiongenetics.com)



ENSURING YOU NEVER WALK ALONE

Providing patients and their providers with expertise in managing critical aspects of the PPP treatment journey

References: **1.** 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. **2.** Cave-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. **3.** Statland JM, Fontaine B, Hanna MG, et al. Review of the diagnosis and treatment of periodic paralysis. *Muscle Nerve.* 2018;57:522-530. **4.** Cannon SC. Channelopathies of skeletal muscle excitability. *Compr Physiol.* 2015;5:761-790. **5.** Hypokalemic Periodic Paralysis. MedlinePlus. Updated March 1, 2020. Accessed November 12, 2024. <https://medlineplus.gov/genetics/condition/hypokalemic-periodic-paralysis/>. **6.** Hyperkalemic Periodic Paralysis. MedlinePlus. Updated February 1, 2019. Accessed November 12, 2024. <https://medlineplus.gov/genetics/condition/hyperkalemic-periodic-paralysis/>. **7.** Paramyotonia Congenita. MedlinePlus. Updated August 1, 2015. Accessed November 12, 2024. <https://medlineplus.gov/genetics/condition/paramyotonia-congenita/>. **8.** Andersen-Tawil Syndrome. MedlinePlus. Updated April 1, 2018. Accessed November 12, 2024. Available at: <https://medlineplus.gov/genetics/condition/andersen-tawil-syndrome/>. **9.** Arya SN. Periodic paralysis. *J Ind Acad Clin Med.* 2002;3:374-382.

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