

PARAMYTONIA  
CONGENITA  
HYPERKALEMIC  
PERIODIC  
PARALYSIS

# Orderable – E-order/Requisition

Turnaround Time: 4 weeks

## Alternate Name(s):

Hyperkalemic Periodic Paralysis Type 1  
PMC

## Specimen:

Whole blood-1 x 4 mL Lavender EDTA top Vacutainer tube

## Collection Information:

Blood samples must be maintained at room temperature.

## Reference Ranges:

See report

## Interpretive Comments:

Paramyotonia congenita (PMC) of Von Eulenburg is an autosomal dominant muscular disease characterized by exercise and cold-induced myotonia and weakness. A number of missense mutations<sup>1,2</sup> in the alpha-subunit of the adult skeletal muscle voltage-gated sodium channel (SCN4A) gene have been identified to cause a spectrum of muscular diseases. These include PMC of Von Eulenburg, PMC without cold paralysis, potassium-aggravating myotonia, and hyperkalemic periodic paralysis (HyperPP). Varying degrees of overlap of the clinical symptoms of PMC and HPP have been recognized, and HyperPP/PMC has been reported in association with eight mutations in the SCN4A gene<sup>3</sup>.

(EX13:I693T, EX13:T704M, EX19:A1156T, EX22:T1313M, EX23:M1360V, EX23:M1370V, EX24:R1448C, EX24:M1592V).

## References

1. Moran. O et al. (1998) *J Bioenerg Biomember* December 30;(6):579-88
2. Yang et al. (1994) *Proc Natl Acad Sci USA* December 20; 91(26):12785-9



**Laboratory:**  
Molecular Diagnostics Lab



**Requisition:**  
[MOLECULAR DIAGNOSTIC  
REQUISITION](#)



**Method of Analysis:**  
DNA Sequence Analysis



**Test Schedule:**  
As required,  
Monday to Friday 0800-  
1600 hours

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3. Brancati et al. (2003) *J Neurol Neurosurg Psychiatry*. 74:1339-41

4. Karin Jurkat-Rott, (2003) *Gene reviews*:

<http://www.genetests.org>

5. <http://blocks.fhcrc.org/sift/SIFT.html>

\*cDNA seq. of SCN4A gene as in gb Accession #NM\_000334. Numbered according to HGVS nomenclature as in den Dunnen et al *Hum Genet* (2001) 109:121-124. initial coding ATG=1

### Storage and Shipment:

Must be received in testing laboratory within 5 days of collection, shipped at room temperature by courier/overnight delivery.