

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^{1,2} 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 gene³.

(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



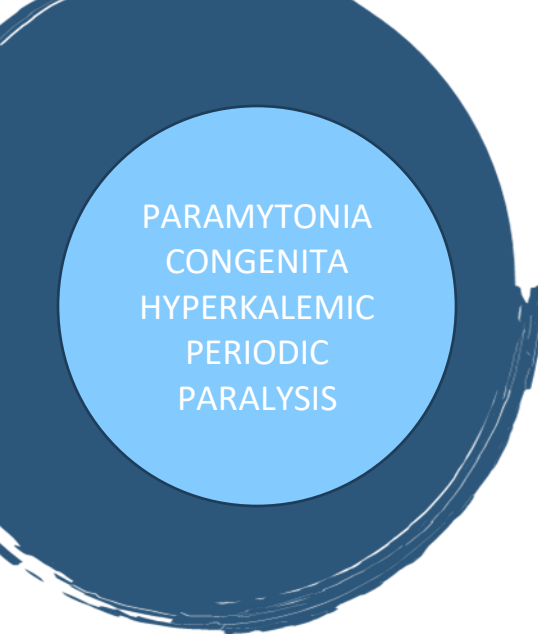
Requisition:
[MOLECULAR
DIAGNOSTICS
REQUISITION](#)



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.