**Orderable – E-order/Requisition**

**Turnaround Time:** 4 weeks

**Alternate Name(s):**

Hyperkalemic Periodic Paralysis Type 1

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

**References**


*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. intial 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.