**cKIT - Mastocytosis**

**Test Code:** 4208  
**Department:** Molecular Oncology

**Test Synonyms:**  
KIT Mutation Screening for Mastocytosis and Mast Cell Leukemia (exon 17)

**CPT Code(s):**  
83890  
83892  
83898  
83905  
83907

**Background:**  
KIT gene mutations are present in the majority cases of mast cell disease/systemic mastocytosis.  
- Adults  
  - D816V mutation is present in 93% of cases  
  - Other mutations are rare (exon 17 D816Y, 815-817 indels; exon 11)  
- Children  
  - D816V mutation is present in 36%  
  - Exon 8 in 18% (mostly del 419)(hereditary in some cases)  
  - Exon 9 in 20% (ITD 502-503; K509I) (hereditary in some cases)  
  - Exon 11 (rare)

**Methodology:**  
DNA is extracted and purified from fresh blood, bone marrow aspirate, paraffin-embedded bone marrow biopsy, clot or other tissue (e.g. skin, GI biopsy).  
- Samples from adult patients are screened for the D816V mutation by real-time allele-specific PCR that includes an internal positive control for DNA quality.  
  - Sensitivity: 1% mutant allele  
- Samples from pediatric patients are screened for the D816V mutation by the above assay; negative samples are further screened for mutations in KIT exons 8, 9 & 11 by standard sequencing.  
  - Sensitivity (exons 8, 9, 11): 20% mutant allele

**Specimen Requirements:**  
- Peripheral blood or bone marrow: 5 – 10 mL in EDTA (purple top) tube. or  
- Paraffin-embedded bone marrow block or  
- 10 unstained sections of tumor (4-5 microns)(15 sections for small biopsies)

A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES. Please include detailed clinical information.

**Test Performed (Days):**  
Mon – Fri

**Turn Around Time:**  
5-7 days
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Shipment Sensitivity Requirements:
Package and ship specimen to remain cold, but not frozen. Ship via overnight express, using the FedEx priority overnight label provided. Contact Client Services at (855) 535-1522 for shipping kits and instructions.

References