Sickle Cell Disease: HbS and HbC Mutations

**Test Code:** HS  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81401 x1

### Condition Description

Sickle cell disease is characterized by intermittent periods of hemolysis and vascular occlusion, causing acute tissue damage resulting in pain and swelling, particularly in the hands and feet.

Chronic symptoms of the disease include:

- Failure to thrive
- Anemia
- Delayed puberty
- Organ dysfunction (nephropathy, cardiomyopathy, retinopathy, and restrictive lung disease).

The disease is caused by mutations in the hemoglobin beta (HBB) gene that result in the formation of the abnormal hemoglobin S (HbS). 60-70% of sickle cell disease is due to inheritance of an HbS allele from each parent (HbSS). The remaining cases of sickle cell disease are due to the co-inheritance of an HbS allele with other abnormal hemoglobin variants, most commonly hemoglobin C (HbC).

Sickle cell disease is inherited in an autosomal recessive pattern. It is more common among individuals of African, Mediterranean, Caribbean, and Hispanic ancestries. Among African-American populations, the carrier frequency of sickle cell trait is approximately 8-10%.

Diagnostic and carrier screening may begin with hemoglobin electrophoresis and CBC, to screen for thalassemias and quantitative hemoglobin variants. For genetic counseling purposes, individuals who are carriers for HbS or HbC, and their partners are offered molecular testing to confirm the specific HBB gene mutation.

Please [click here](#) for the GeneClinics summary on this condition.

### Genes

**HBB**

### Indications

This test is indicated for:

- Individuals with symptoms of sickle cell or sickle-C disease.
- Carrier screening in persons of African, Mediterranean, Caribbean, and Hispanic ancestries.
- Testing for carrier status in family members of an individual with sickle cell disease.

### Methodology

Presence or absence of the hemoglobin S and C mutations mutations are detected by pyrosequencing analysis.

### Detection

This assay will detect over 99% of HbS and HbC alleles.

### Reference Range

Qualitative assay.

### Specimen Requirements

**Type:** Whole Blood

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

### Related Tests

- Prenatal custom diagnostics (NW) for sickle cell disease may be available to couples who are confirmed carriers of the sickle cell trait. Please...
contact the laboratory genetic counselor to arrange testing prior to collecting a prenatal specimen.