

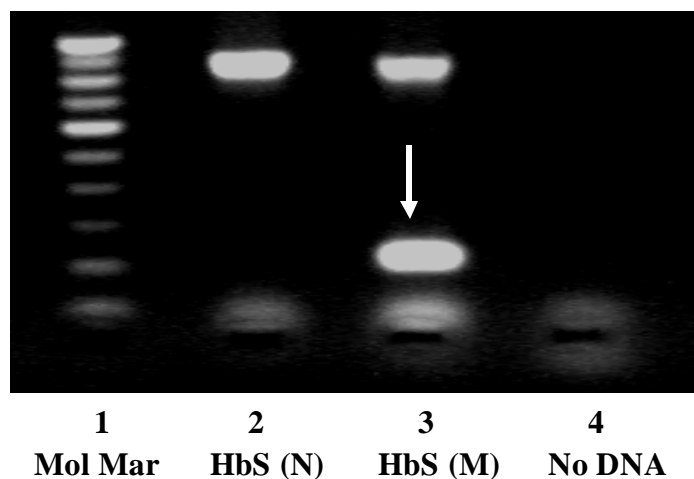
Prenatal Diagnosis of Sickle Cell Anemia

A non-consanguineous couple with history of a child with sickle cell anemia attended genetic counseling at GeneTech. The wife was 10 weeks pregnant at the time of counseling and the couple was concerned about the outcome of the pregnancy. There were incidents of sickle cell anemia in the extended family and both the parents were known carriers of sickle cell trait.

In sickle cell anemia, the red blood cells become hard, sticky, and shaped like sickles or crescents. The disease is characterized by variable degrees of haemolysis and intermittent episodes of vascular occlusion resulting in tissue ischemia and acute and chronic organ dysfunction. Consequences of haemolysis include chronic anemia, jaundice, predisposition to aplastic crisis, and delayed growth and sexual maturation. Sickle hemoglobin (HbS) results from a single point mutation in which the codon determining the amino acid at position $\beta 6$ has changed from GAG coding for glutamic acid to GTG coding for valine. Recurrence risk for carrier parents is 25% for all future pregnancies.

Prenatal diagnosis by chorionic villus sampling was advised. Molecular analysis for β -Thalassemia mutations on CVS sample revealed HbS mutation in homozygous condition indicating that the fetus is affected with Sickle cell anemia. The result was communicated to the couple within 1 week.

The following picture of agarose gel shows DNA stained with ethidium bromide. Column 1 is molecular marker reference with known molecular weight. Column 2 is DNA amplified with normal primers (normal Hb) and shows no amplification. Column 3 is DNA amplified with mutation specific primers (mutation HbS) showing amplification. This indicates that both the chromosomes have mutated gene and normal sequence is absent. Column 4 represents negative control (No DNA) for quality assurance.



For further details on Sickle Cell /Thalassemia testing call our 24 hr customer care # **98480-41127**