

Sickle Cell Anemia

Mutation Detection Test



Early Diagnosis • Detect precise cause of disease
Reduce chances of incorrect diagnosis

Sickle Cell Anemia

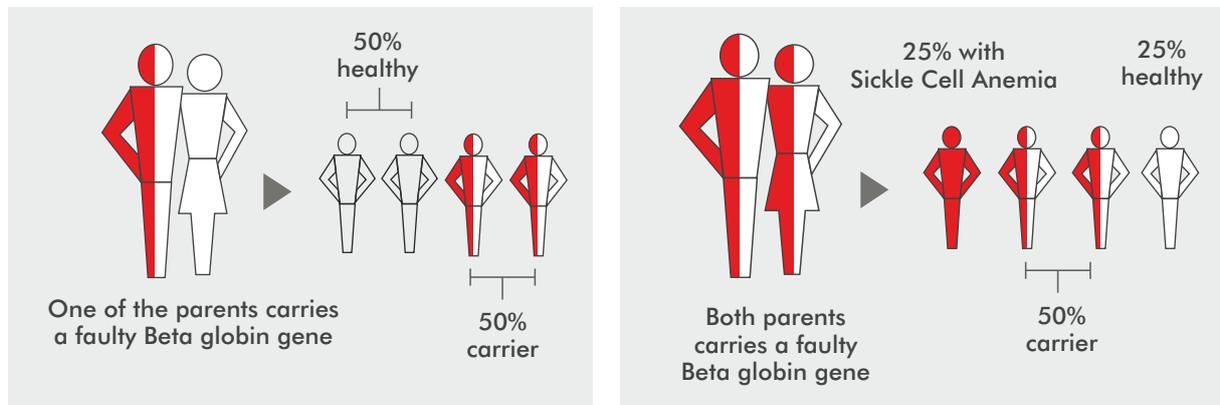
In India there are about 270 million carriers of sickle cell anemia and/or thalassemia (WHO 1994). Each year about 40000 individuals are born with serious hemoglobinopathies due to this mutation.

Sickle Cell Anemia is a hereditary genetic disorder in which the body makes an abnormal form of hemoglobin, a protein in red blood cells that carries oxygen. HBB gene is responsible for production of beta-globin protein of the hemoglobin molecule. It is located on chromosome 11p15.5. Different kind of mutations in HBB gene are responsible for formation of faulty hemoglobin. One particular HBB gene mutation produces an abnormal version of beta-globin known as hemoglobin S (HbS). HbS has a substitution of valine for glutamic acid at the sixth position of the beta-globin chain from normal. People with this disorder have a deformed hemoglobin in blood called hemoglobin S, which can distort red blood cells into a sickle or crescent shape. The sickle-shaped red blood cells are incapable of carrying oxygen and die prematurely, which often lead to severe anemia. Sickle Cell Anemia can cause shortness of breath, fatigue, and delayed growth and development in children.

Importance of Screening

Sickle Cell Anemia is a genetic abnormality which is inherited from parent to progeny and follows autosomal recessive pattern of inheritance. If both the parents are carriers, there are 25% chances of their children carrying the faulty gene. Therefore, it is strongly recommended to undergo genetic testing, to avoid Sickle Cell Anemia condition in children. The couple who are carrier for sickle cell trait should definitely undergo genetic testing for Sickle Cell Anemia to find out whether their future child can have this disease or not. Pre-natal screening* of pregnant ladies at earlier stages is required if any of the parents are identified as carrier after conception. Screening of Sickle Cell Anemia at early age helps in better treatment and management of disease.

(* Please discuss it with your Physician or Genetic Counsellor)



Why go for molecular testing ?

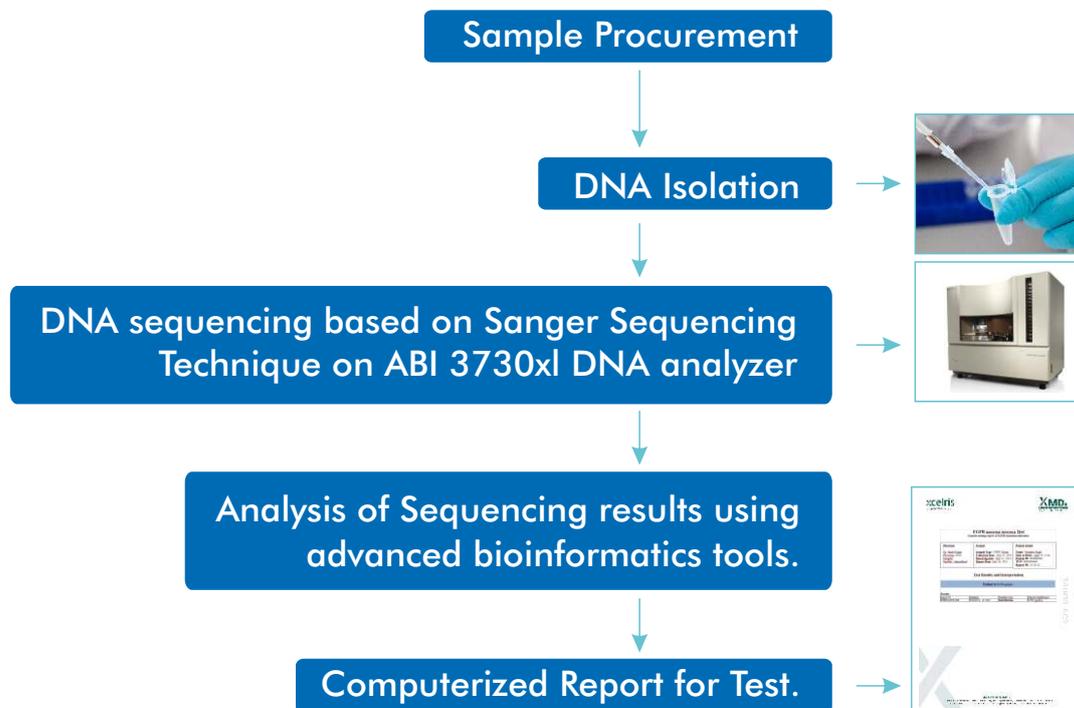
High Specificity and Precision: Molecular testing can detect and measure presence of minimal residual gene mutation or protein associated with disease condition which cannot be done by conventional biochemical testing, thus reveals precise cause of disease.

High coverage and accuracy: Gene sequencing technique detects all possible mutations in gene which can find out molecular genetic basis of complex diseases and nullifies the chances of false positive or false negative results, thereby reducing chances of incorrect diagnosis.

Early diagnosis and efficacy: Molecular diagnosis can detect disease at early stage which helps clinicians to monitor the progression of disease and determine effective drug therapy for the patient.

Methodology for Testing

Gene specific PCR coupled with Sanger Sequencing is presently used to identify the gene mutations in HBB gene.



Sample Requirement

A. **Blood Sample:** 3ml-5ml blood in EDTA Vacutainer. **(Product Code: HDSC01)**

B. **Saliva Sample:** Min 2ml Saliva Sample in pre-designed kits provided by Xcelris Labs. **(Product Code: HDSC02)**

C. **Amniotic Fluid:** Min.20ml of Amniotic fluid / CVS in transfer buffer in a sterile container along with Blood sample of both parents in EDTA Vacutainer. **(Product Code: HDSC03)**

Specimen Handling:

Do not heat, freeze or centrifuge sample before shipment. Refrigerate sample until shipment of the same.

Transport:

Blood and CVS Sample: Send the sample at 2 to 8°C temperature with overnight delivery for receipt Monday through Friday within 24 hours of collection.

Saliva Sample: Send the sample at room temperature to Xcelris Labs from Monday to Friday by overnight delivery service within 24 hours of collection.

Unacceptable conditions:

Contaminated or deteriorated sample from which DNA cannot be extracted and mislabeled specimens will not be accepted, it will be liable for re-sampling.

Turn Around Time :

7 to 10 working days after receiving of sample at Xcelris Labs.

How to Order Test ? :

You can order test with following product code

HDSC01 (Blood Based Test)	HDSC02 (Saliva Based Test)	HDSC03 (Prenatal Test)
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*Pre-natal Sickle Cell Anemia test can be ordered by Clinicians only. Kindly consult your Physician or Genetic Counselor.

Contact Details

Name : Mobile.:

About Xcelris

Xcelris is one of the leading genomic research organizations and service provider offering cutting edge solutions to the life science industry and research institutions.

Xcelris Molecular Diagnostics (XMDx) offers clinical testing services based on Real time PCR, Sequencing and Microarray technology using latest state of art platforms. Xcelris Labs provides genetic testing for cancer, inherited disorders, drug response screening and molecular testing for infectious diseases.

At Xcelris, we believe that Next Generation Sequencing Technology will be a break through in the diagnostic segments by which clinicians will be able to track diseases at early stages, making clinical management more effective and easy.

