**Beta Thalassemia**

**Mutation Detection Test**

**Beta Thalassemia Mutation**

Thalassemia is a hereditary genetic disorder in which the body makes an abnormal form of hemoglobin, a protein in red blood cells that carries oxygen. Thalassemia occurs when our body is not able to produce sufficient amount of hemoglobin. It is caused by mutations in gene encoding Beta globin protein of the hemoglobin.

In such case excessive destruction of red blood cells is observed which results in severe anemia. Children affected with Beta Thalassemia exhibits the symptoms of anemia, poor growth and skeletal abnormalities during infancy. Patients with Thalassemia major can develop life threatening anemia and require regular blood transfusions for survival and may develop significant implications including iron overload, bone deformities, cardiovascular failure, spleen enlargement and even premature death.

**Early Diagnosis • Detect precise cause of disease**

**Reduce chances of incorrect diagnosis**

Xcelris Molecular Diagnostics

Pragnosis | Diagnosis | Theranosis
HBB gene is responsible for production of Beta globin protein of the hemoglobin molecule. It is located on chromosome 11p15.5. Mutations in HBB gene leads to Beta Thalassemia. This disorder is of two different types- Thalassemia Major and Thalassemia Minor, depending upon the type of mutations present in patients. In Thalassemia Minor patients there is reduced ability to produce Beta-globin protein and reduced functional capacity of hemoglobin. In Thalassemia Major patients Beta-globin is not produced and has dysfunctional hemoglobin.

Prevelance of Beta-Thalassemia in India
Beta Thalassemia is one of the most common blood disorders observed in Indian Subcontinent. In India 1 out of 14 people are reported as carrier for different type of Thalassemia. Each year about 40,000 infants are born with serious hemoglobinopathies and carrier frequency is 270 million. There are 10 common mutations including base substitutions, insertions and deletions which have been observed in Indian population. Four of these are classified as $\beta^+$ and six are classified as $\beta^0$.

Importance of Screening
Thalassemia is a genetic disorder which can be inherited from parents to their progeny. It follows autosomal recessive pattern of inheritance. If both the parents are carrier, there are 25% chances of their children carrying the faulty gene. Therefore, it is strongly recommended to undergo genetic testing for Beta Thalassemia to avoid serious consequences in newborn children.

One should undergo Beta Thalassemia genetic testing in the following conditions:
A. Marriage: Premarital test using blood/saliva sample is available to find out whether any one of the partner or both partner are Beta Thalassemia carriers.

B. Family planning: Couple who are planning a family should undergo genetic test to identify Beta Thalassemia carrier status in any of the parent. This test helps to find possibility of children carrying faulty thalassemia gene.

C. Pregnancy: In pregnancy the genetic test is performed with amniotic fluid or CVS sample to screen foetus for disease. This test identifies that whether foetus will have Beta Thalassemia or not.

Technology used - Sequencing Technology
Based on available genomic data, more than one kind of mutations are observed in Indian population. Conventional methods using commercial PCR kits, are unable to detect multiple mutations in Beta Thalassemia gene. Sequencing technology involves full gene sequencing with careful reading of whole HBB gene to detect mutation in the patient DNA sample. If more than one kind of mutation are prevalent within single DNA sample it can be identified with sequencing technology.
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.

Types of mutation covered in genetic testing

Many different type of mutations prevail among Indian population and are distributed across various geographical locations. We identify this mutation using latest sequencing technology platform at Xcelris Labs.

### Test Methodology

1. **Sample Procurement**
2. **DNA Isolation**
3. **DNA sequencing based on Sanger Sequencing Technique on ABI 3730xl DNA analyzer**
4. **Analysis of Sequencing results using advanced bioinformatics tools.**
5. **Computerized Report for Test.**

### Test Report Interpretation

<table>
<thead>
<tr>
<th>Mutation type</th>
<th>Possible Genotype*</th>
<th>Clinical Significance</th>
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<tbody>
<tr>
<td>β⁺ type</td>
<td>β⁺ β⁺</td>
<td>Thalassemia Major</td>
</tr>
<tr>
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<td>β⁺ β⁻</td>
<td>Thalassemia Major</td>
</tr>
<tr>
<td></td>
<td>β⁺ β</td>
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Sample Requirement

A. **Blood Sample:** 3ml-5ml blood in EDTA Vacutainer. *(Product Code: HDBT01)*

B. **Saliva Sample:** Minimum 2ml Saliva Sample in pre-designed kits provided by Xcelris Labs. *(Product Code: HDBT02)*

C. **Amniotic Fluid:** Minimum 20ml of Amniotic fluid / CVS in transfer buffer in a sterile container along with blood sample of both parents in EDTA Vacutainer. *(Product Code: HDBT03)*

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 2-8°C temperature with overnight delivery for receipt Monday to 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:** 7-10 working days after receiving of sample at Xcelris Labs.

**How to Order Test?** You can order test with following product code

<table>
<thead>
<tr>
<th>Product Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>HDBT01</td>
<td>(Blood Based Test)</td>
</tr>
<tr>
<td>HDBT02</td>
<td>(Saliva Based Test)</td>
</tr>
<tr>
<td>HDBT03</td>
<td>(Prenatal Test)</td>
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</tbody>
</table>

Contact our Local Representative or email us at diagnostics@xcelrislabs.com

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.