Lactose intolerance is observed worldwide affecting all kinds of people. A significant percentage of the human population is affected by hypolactasia (80-100% in Asia, Africa) - a genetic mutation condition. Lactose intolerance in Indian population is mostly misunderstood as milk product allergy. Primary or neo-natal lactase deficiency is a genetic gastrointestinal disorder in which body is unable to digest lactose – a sugar found in milk and dairy products. Medically this condition is called Hypolactasia or lactase deficiency. The affected individuals show very low level of lactase enzyme in their gastrointestinal tract. Ingestion of milk may lead to abdominal cramps, bloating, distension, flatulence and diarrhea. Symptoms of lactose intolerance are sometimes intolerable by the patient.

The LCT gene is responsible for the production of lactase. LCT gene expression is controlled by a DNA sequence called regulatory element, which is located within a nearby gene called MCM6 (Mini chromosome maintenance gene). This gene is located on chromosome 2, upstream from the lactase gene (LCT), encoding for lactose phlorizin hydrolase (LPH), converting lactose into glucose and galactose. Mutation in MCM6 gene is responsible for lactose intolerance.

**Importance of Screening**

Lactose intolerance is a genetic mutation which is passed on from one generation to another. This condition is inherited in autosomal recessive pattern, where the parents who carry the faulty gene have chances to pass this gene to their children. Screening of lactose intolerance is important because if this condition is inherited in the children, then they will not be able to digest milk – a prime source of nutrition. So early detection of this gene mutation is very important. For adults, if screening is done at earlier stage treatment, management and diet regulation becomes easy. A genetic test for lactose intolerance can help you understand if you have the genetic variation that means you produce less lactase as you get older.
**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.

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**Methodology for Testing**

1. **Sample Procurement**
2. **DNA Isolation**
3. **Sequencing of DNA**
4. Analysis of DNA sequencing data using advance bioinformatics tools
5. **Test Report**

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**Sample Requirement:**

**Saliva Sample:** Min 2ml Saliva Sample in pre-designed kits provided by Xcelris Labs. *(Product Code: HDLI02)*

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

**Transport:**

**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:** 6 working days after receiving of sample at Xcelris Labs.

**How to Order Test?** You can order test with following product code **HDLI02 (Saliva Based Test)**

Contact our local representative or email us at diagnostics@xcelrislabs.com