Sonic Genetics

Information for Doctors

# Lactose intolerance and genetic testing

Sonic Genetics offers a genetic test to assist in the diagnosis of lactose intolerance in both children and adults.

## Key points

- Lactose intolerance affects approximately 75% of the global population.
- Genetic testing can confirm lactose tolerance (also referred to as lactase persistence).
- The test differentiates between primary lactose intolerance, due to lactase deficiency, and secondary causes of lactose intolerance, due to other more serious conditions that affect the small bowel.
- The test is not affected by intercurrent illness and can be performed non-invasively on patients of all ages.
- The test only needs to be performed once during a person's lifetime.

## How does the test work?

Testing is available to detect the genetic variant that accounts for close to 100% of lactase persistence in Europeans (the enhancer variant upstream of the LCT gene commonly known as "-13910C>T"). Three other genetic variants that have a similar effect and are more common in non-European populations are also detectable. These variants are thought to act as enhancers of the lactase gene that in turn stimulates lactase production.

The clinical utility of this test is its application in excluding primary lactose intolerance as the underlying cause of gastrointestinal symptoms, by detecting a genetic variant that results in lactase persistence. It may also assist in distinguishing between primary lactose intolerance and lactose intolerance secondary to other conditions that affect the small bowel, such as gastroenteritis, inflammatory bowel disease and coeliac disease.

## What causes lactose intolerance?

Lactose is the major carbohydrate in mammalian milk. Lactose intolerance is caused by deficiency of lactase, the enzyme required for digestion of lactose. Symptoms include abdominal pain, diarrhoea, nausea, flatulence and/or bloating, following the consumption of lactose-containing foods.

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Testing is performed in Australia at one of our NATA-accredited laboratories.

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## Who is affected by lactose intolerance?

After infancy, approximately 75% of the global population lose the ability to digest lactose, due to a deficiency in lactase, referred to as primary lactose intolerance. The remainder of people maintain their tolerance for lactose-containing foods because of genetic variants that enable continued production of lactase, referred to as lactase persistence.

The prevalence of primary lactose intolerance varies significantly with ethnic background. Lactose intolerance is uncommon in populations that consume large amounts of dairy, for example, northern Europeans (as low as 10%), but is frequent in other populations (as high as 100% in Asiatic countries). It is hypothesised that this is the result of selective genetic advantage; populations that have historically been dependent on dairy food sources for nutrition have survived by having genetic variants that allow tolerance for lactose.

## Other testing alternatives

Currently, testing for lactose intolerance can also be performed by a hydrogen breath test with lactose load, or by measurement of intestinal lactase enzyme activity in a biopsy obtained during endoscopy. These tests may give a false-positive result when lactase levels have been affected by a recent viral illness or coeliac disease. These procedures are also not suitable for testing children younger than seven years old. Genotyping is not affected by intercurrent illness and can be performed non-invasively on patients of all ages.

## Genetic testing limitations

Genotyping will not identify very rare genetic variants associated with persisting lactase activity, and therefore the absence of a variant should only be used to support a diagnosis of lactose intolerance along with other clinical and laboratory findings.

## Arranging a test

1. Complete a standard pathology request form to refer your patient for 'lactase persistence' or 'lactose intolerance genetic testing'.
2. Your patient can have a blood sample or buccal swab taken at any of our pathology collection centres. No special preparation or booking is necessary.
3. The sample is tested at one of our NATA-accredited laboratories supervised by a genetic pathologist.
4. Results are provided within 5 business days of the laboratory receiving the sample.

## Cost

Please to refer to our website for current pricing, sonicgenetics.com.au.

Medicare does not cover the cost of this test and your patient will receive an invoice for $75.*


References

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For further information, please refer to our website, sonicgenetics.com.au or call us on 1800 010 447


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