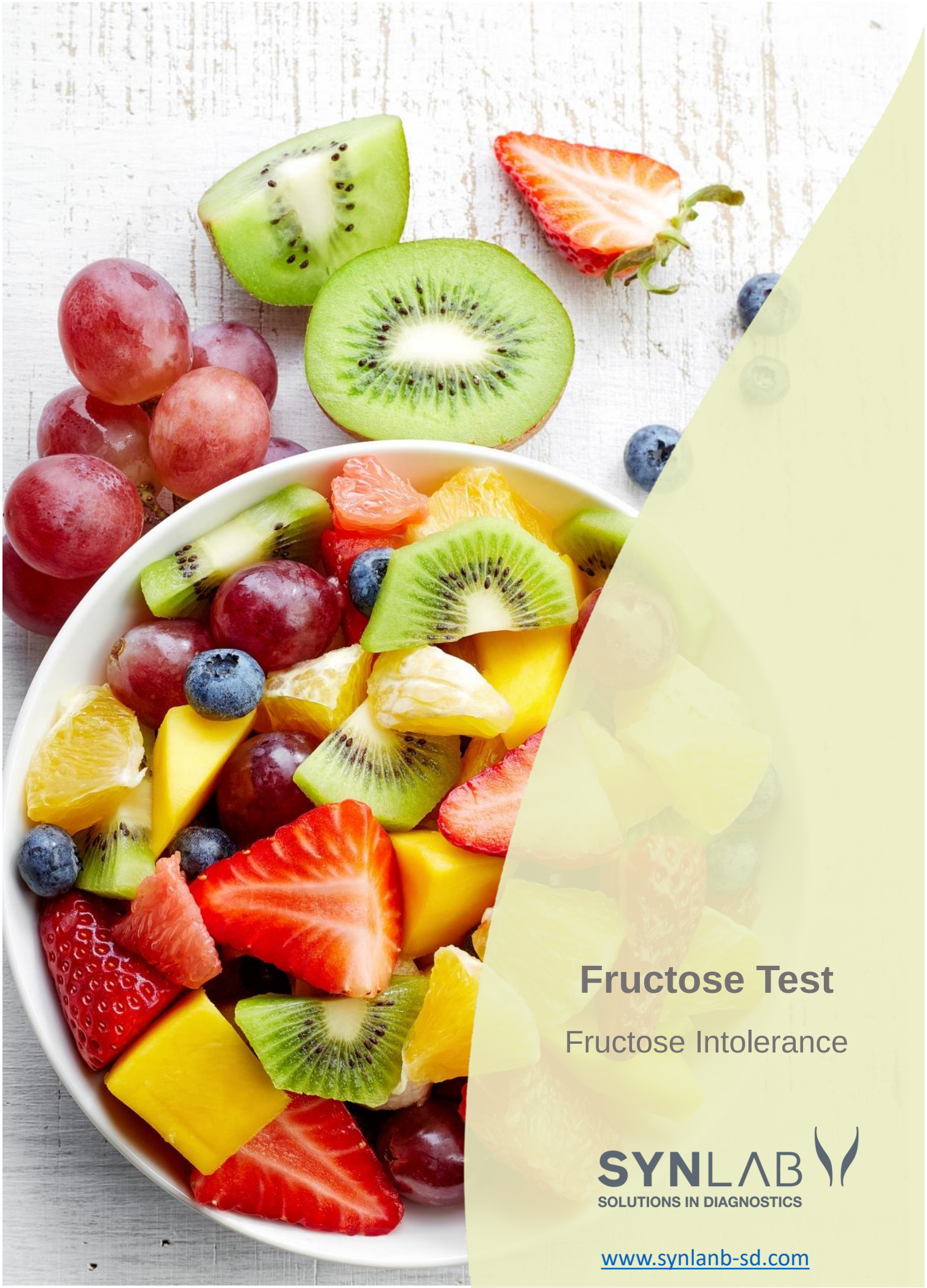

Fructose Test

Fructose Intolerance

SYNLAB
SOLUTIONS IN DIAGNOSTICS

www.synlanb-sd.com

Why undergoing this examination?

Hereditary fructose intolerance (HFI) is a disorder of fructose metabolism, resulting from a deficiency in the activity of the enzyme aldolase B (fructose-1-phosphate aldolase). This enzymatic defect prevents the transformation of fructose into its various metabolites, resulting in inhibition of glucose synthesis and a decrease in adenosine triphosphate (ATP). As a consequence, various metabolic alterations such as hypoglycemia, lactic acidemia, hypophosphatemia, hyperuricemia, hypermagnesemia, and hyperalaninemia are observed, along with clinical symptoms such as nausea, vomiting, and abdominal pain. HFI is a genetic disease inherited in an autosomal recessive manner, resulting from mutations in the *ALDOB* gene.

What is the exam?

The **Fructose Test** involves the evaluation of the three recurrent mutations in the *ALDOB* gene (A149P, A174D, N334K), responsible for 90% of HFI cases in European patients. Alternatively to the breath test, the analysis is performed from a blood sample, avoiding direct patient exposure to substances.

For whom is it indicated?

- Children with clinical symptoms and nutritional and/or family characteristics suggestive of HFI.
- Close relatives of patients with HFI.
- Clinical suspicion of fructose malabsorption.

Technology

Sanger Sequencing

Advantages

SYNLAB GROUP

Guaranteed by the experience of the absolute European leader in laboratory diagnostics.

COMPLETE

- Report with objective results and detailed description.

Extra Information

DOCUMENTATION - Available on the SYNLAB Direct for clients

- Informed consent;
- Clinical questionnaire.

PREPARATION

- Fasting is not necessary for the exam.

Delivery Time

30 business days

Sample Type

5 mL of whole blood in EDTA