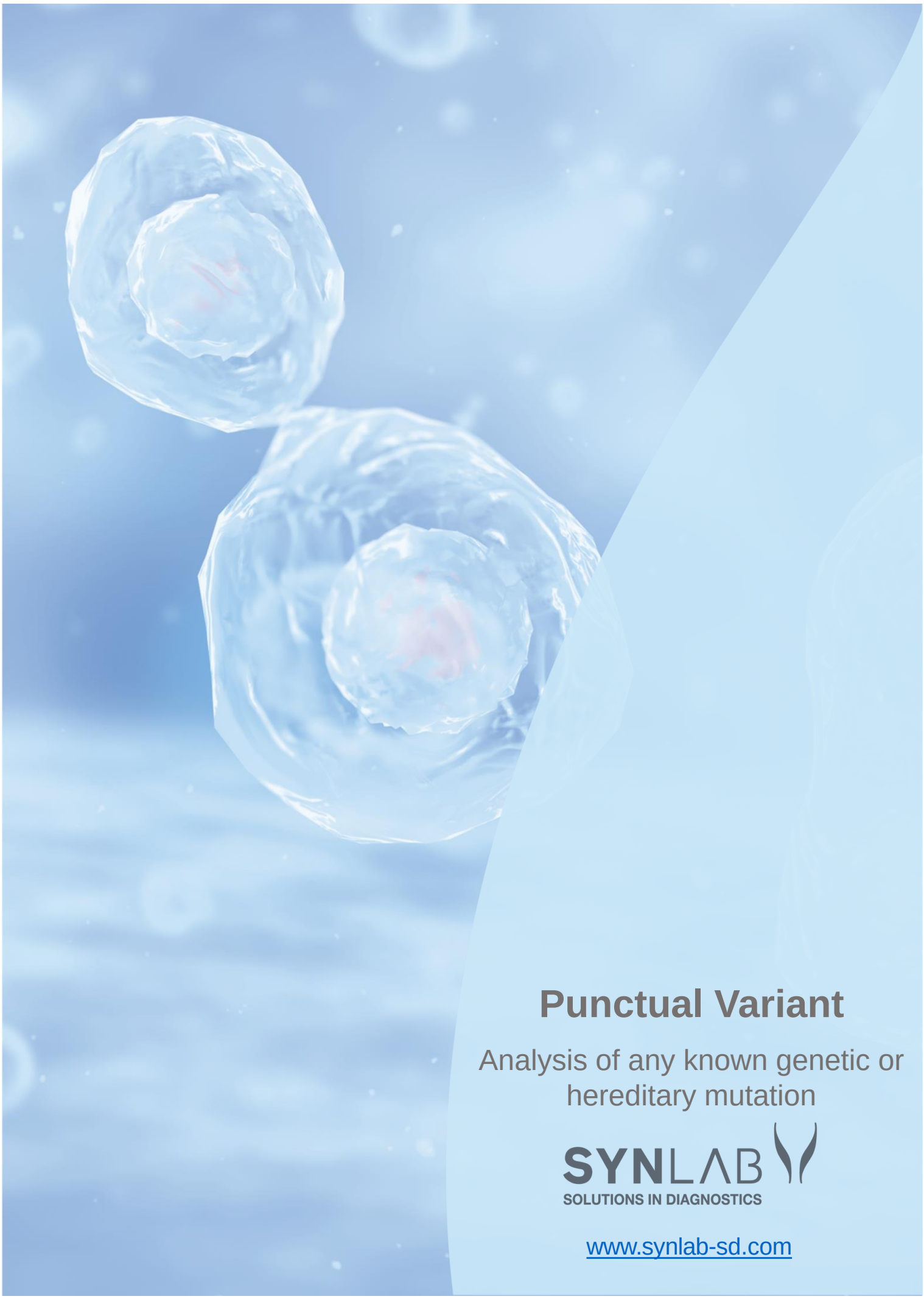

Punctual Variant

Analysis of any known genetic or hereditary mutation

SYNLAB
SOLUTIONS IN DIAGNOSTICS

www.synlab-sd.com

Why undergoing this examination?

The term mutation refers to a change in the DNA nucleotide sequence that can result in altered proteins or gene expression. A DNA change can either enhance the organism or lead to genetic disorders, known as pathogenic mutations.

What is the exam?

The **Ponctual Variant** test involves the analysis of any genetic alteration previorsly reported in a family member.

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For whom is it indicated?

- Individuals with a family history of hereditary cancer who want to confirm if they have the same mutation identified in the family.

- Patients with a family history of a genetic syndrome who want to confirm if they have the same mutation identified in the family.

Technology

Sanger Sequencing

Advantages

SYNLAB GROUP

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

COMPLETE

- Complete genetic study of genes related to hereditary gastrointestinal cancer;
- Any pathogenic or uncertain significance variant is confirmed throrgh a new DNA extraction and Sanger sequencing.

Extra Information

DOCUMENTATION – Available on the SYNLAB Direct for clients

- Informed Consent;
- Clinical Questionnaire;
- Family Medical History;
- Medical Request.

PREPARATION

- Fasting is not necessary for the exam.

Delivery Time

45 business days

Sample Type

5 mL of total blood in EDTA