Good clinical performance of the LAMP Human Lactose Intolerance KIT on buccal-swabs as an alternative for hydrogen breath test

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Introduction

Lactose intolerance, the impaired ability to digest lactose in adulthood, is regulated by MCM6 in which several variants have been described that give rise to a lactase-persistent phenotype. The current hydrogen breath test is not patient friendly. Therefore, we aimed to test a rapid, reliable and patient-friendly alternative.

Methods

A cohort of 40 patients were tested for lactose intolerance via a hydrogen breath test and, simultaneously, a buccal-swab and EDTA-blood sample were collected. The EDTA-blood samples were tested by an external laboratory for the four most prevalent lactose-persistent variants, -13910C>T, -13915T>G, -13907C>G and -13913T>C using a DNA-extraction, PCR and melting curve analysis. In our laboratory, the blood- and swab-samples were tested using the LAMP Human Lactose Intolerance KIT (LaCAR-MDx) on a LC480II (Roche). Samples were subjected to a short lysis, directly followed by LAMP for the detection of the four variants.

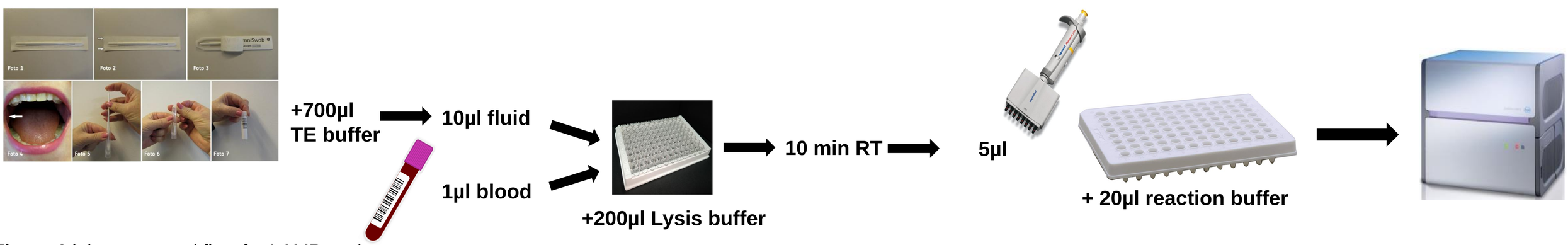

Figure 1:laboratory workflow for LAMP testing

Results

The three DNA-tests using blood and buccal swabs showed a 100% concordance (table 1). All patients with the wildtype variant had a positive hydrogen breath test, indicating impaired ability to digest lactose. All 17 -13910C>T homozygous patients and 14 out of 15 -13910C>T heterozygous patients where negative in the breath test. One patient had a -13910C>T heterozygous variant in the DNA tests, but had a positive breath test. The blood sample was send out for extensive DNA testing confirming our genotyping findings (table 2). Most likely this patient has a secondary lactose intolerance.

Genotype	Reference blood test	blood LAMP	SWAB LAMP
13910CC (wildtype)	8	8	8
13910CT (heterozygous)	15	15	15
13910TT (homozygous)	17	17	17

Table 1:genotypes found in the test cohort

	breath test	
Genotype	negatieve	positive
13910CC (wildtype)	0	8
13910CT (heterozygous)	14	1
13910TT (homozygous)	17	0

Genotype confirmed by reference laboratory

Table 2:Genotype compaired to breath test

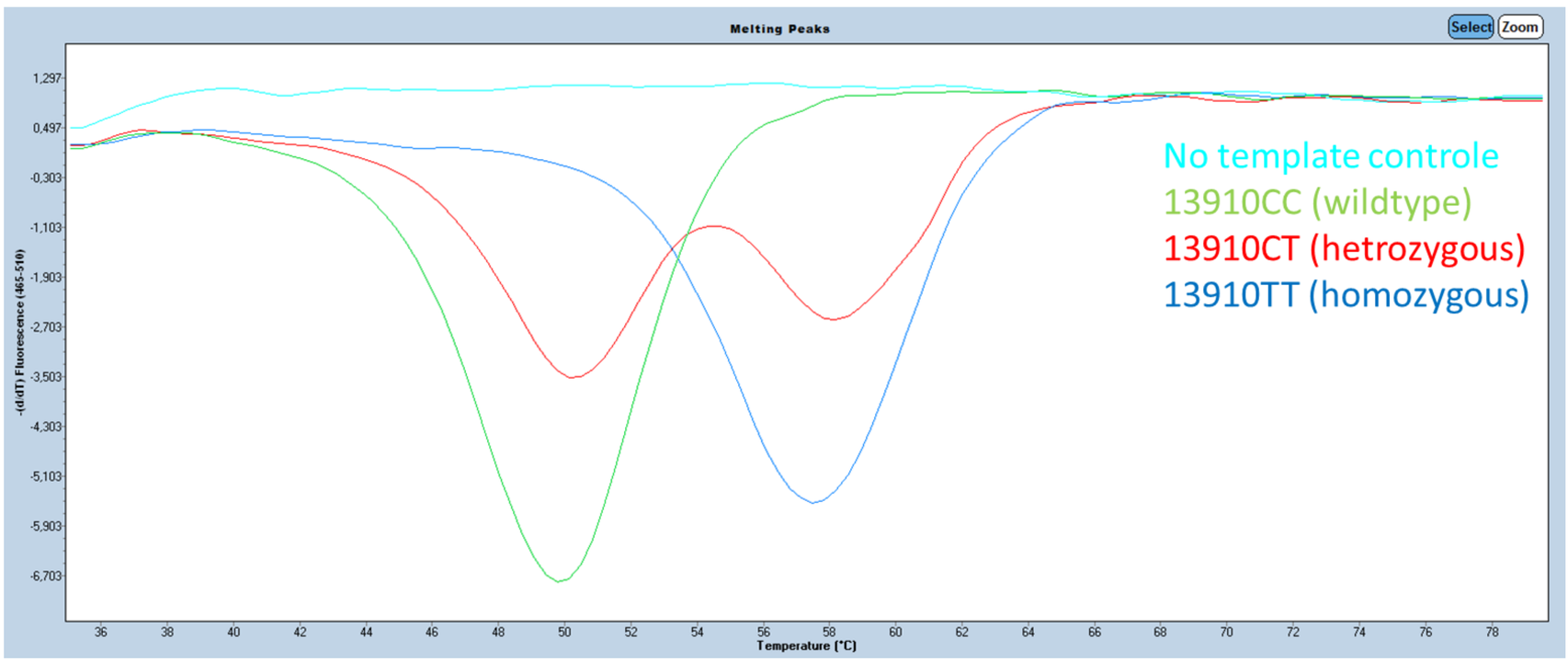

Figure 2:representive example of LAMP result on LC480II

Conclusion

The LAMP Human Lactose Intolerance KIT on buccal-swabs or EDTA-blood is an (at least) equally well performing and a patient friendly alternative for the hydrogen breath test. It is easy to use in a laboratory setting and gives fast results without elaborate DNA extraction.

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