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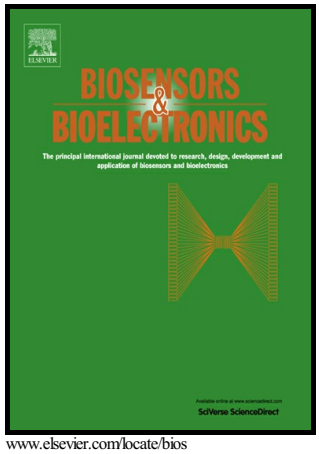

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Polymorphism genotyping based on loop-mediated isothermal amplification and smartphone detection

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ABSTRACT

The genotyping of a single-nucleotide polymorphism (SNP) is addressed through methods based on loop-mediated isothermal amplification (LAMP) combined with user-friendly optical read-outs to cover the current demand for point-of-care DNA biomarker detection. The modification of primer design and reaction composition improved the assay selectivity yielding allele-specific results and reducing false-positive frequency. Furthermore, the reduced cost, ease of use and effectiveness of colorimetric detection (solution and hybridization chip formats) were availed for the image capture by a smartphone, reching high sensitivity. In order to evaluate their discriminating capacities, LAMP-based methods were applied to human samples to genotype an SNP biomarker (rs1954787) located in the *GRIK4* gene and related to the treatment response to anti-depressants drugs. Sensitive (limit of detection: 100 genomic DNA copies), reproducible (<15% error), fast (around 70 min) and low-cost assays were accomplished. Patient subgroups were correctly discriminated, agreeing with reference sequencing techniques. The achieved analytical performances using the developed amplification-detection principles confirmed the approach potential for point-of-care optical DNA testing.

Graphical abstract