

Original Article

Novel detection of IVS I-1 G>T mutation on β -globin gene

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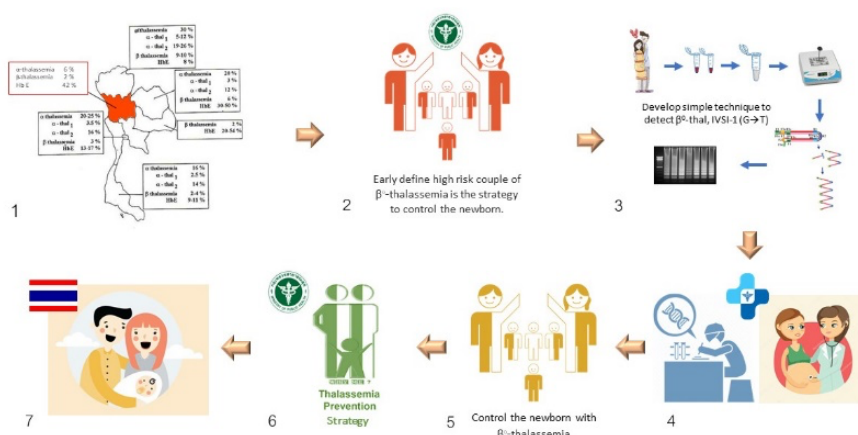
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Abstract: Mutation at IVS I-1, G>T on β -globin gene is one of the β^0 -thalassemia, caused severe clinical symptoms found in Thailand. Detection of this β^0 -globin gene mutation in the carriers to define the risk couple is a strategy to control newborn with β^0 -thalassemia. We developed the reliable Loop-mediated isothermal amplification (LAMP) assay to detect this mutation in one-tube isothermal amplification. Firstly, we designed 4 sets of LAMP primers specific to β^0 -globin gene, IVS I-1, G>T mutation, then optimized the amplification condition and the DNA amount; and finally applied on wildtype and heterozygote for IVS I-1, G>T mutation. The results showed that LAMP primer set 4 was successfully differentiated between normal allele and IVS I-1, G>T mutation allele with no cross amplification. This assay could detect mutation allele using 5-10 ng DNA and completed reaction in a single temperature within 50 min. The result was in accordance with the conventional technique, multiplex Amplification Refractory Mutation System (multiplex ARMS) technique. Moreover, we found that heating block, an inexpensive equipment could replace a thermo cycler machine, to incubate LAMP reaction with one temperature. This simple, rapid and cost-effectiveness LAMP assay could be useful to identify IVS I-1, G>T mutation and other β^0 -globin gene mutations in primary hospitals. Further studies are needed to develop the LAMP assay to identify other β^0 -globin gene mutations mostly found in Thailand. This assay could support the national program to control the incidence of β^0 -thalassemia in Thailand.

Graphical abstract:



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Keywords: β^0 -thalassemia, IVS I-1 detection, LAMP

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