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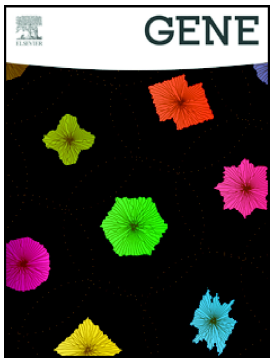


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# The Sensitive Detection of IVSII-1(G > A) Mutation in Beta Globin Gene Using a Nano-based Ligation Genotyping System

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## Abstract

Beta-thalassemia (β-thalassemia) is a globally genetic diseases, and is most prevalent in the Middle East, particularly in Iran. Carrier detection and prenatal diagnosis are the best ways to manageing it, and to prevent new community cases from emerging. We report on a simple method for rapid detection of the worst β-thalassemia point mutation in Iran (IVS-II-1 G>A), using a nano-based ligation assay, this was performed using probes with labeled magnetic nanoparticles and quantum dots. After optimizing the technique, 50 DNA samples were genotyped with this method. We found a frequency of 72% for IVSII-1 (G>A) mutation (42% heterozygote, and 30% mutant homozygote) with a highly sensitive nano-based ligation genotyping system, offering excellent sensitivity and specificity for point mutation detection; it has been demonstrated to be anaccurate, sensitive, cost-effective, and rapid technique for single nucleotide polymorphism (SNP) genotyping.

**Key words:** Beta-thalassemia; beta globin; IVSII-1 (G>A); nano-based ligation assay.

## 1. Introduction

Haemoglobinopathies are the diverse group of the inherited recessive disorders which results in the abnormal structure and reduced (or absent) synthesis of the globin chains in the hemoglobin molecule. Two common types of thalassemia (alpha and beta-thalassemia)

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