



Medical Genetics Laboratory of Dr. Zeinali

The significant role of STR markers in complex diagnosis of PND or PGD of β thalassemia

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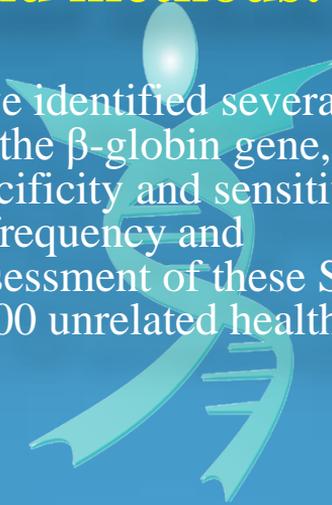
Introduction:

β -thalassemia is the most common hematological disorder worldwide. Because of the high carrier rate of beta globin gene mutations in Iran, prenatal diagnosis (**PNDs**) or Pre-implantation Genetic Diagnosis (**PGD**) are the favored procedures to prevent birth of new β -thalassemia cases.

Haplotyping, using STRs (**Short Tandem Repeat**) markers linked to β -globin gene, would add accuracy to any PND and is essential for every PGD for β -thalassemia.

Materials and methods:

In this research, we identified several novel STR markers linked to the β -globin gene, with the aim of increasing the specificity and sensitivity of the diagnosis. Allele frequency and heterozygosity assessment of these STR markers were studied on 100 unrelated healthy Iranian individuals.

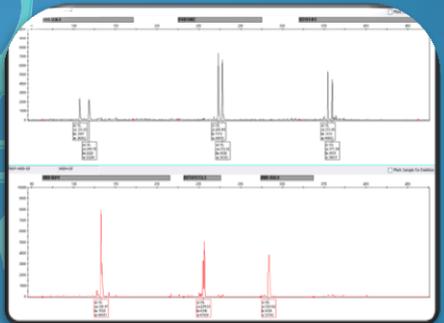
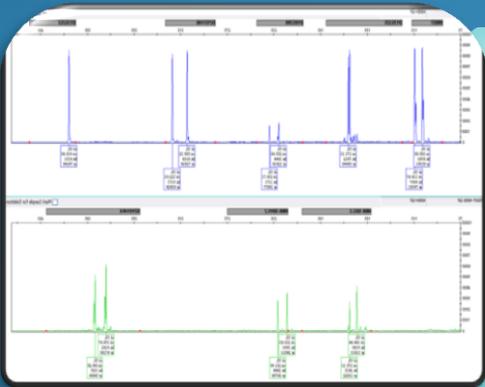




Result

In total, **97** alleles were detected for ten STR loci. The observed range of allele frequencies was from **0.7%** to **38 %**. The frequencies of genotype distributions for all ten STRs were found to be **$P \geq 0.1876$** which is in agreement with the Hardy–Weinberg equilibrium.

Genotyping of all couples were determined by ARMS, MLPA or Sanger sequencing methods. Molecular findings were compared with using linkage analysis based on family studies. Also, PGD was applied for 14 couples at risk of transmitting β -thalassemia to their offspring.





Discussion:

We found these markers can be easily applied for PGD, PND of beta thalassemia. This panel increases the **specificity** and **sensitivity** of the diagnosis.

