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Detection of β-thalassemia IVSI-110 mutation by using piezoelectric biosensor for non-invasive prenatal diagnosis

Umut Kokbas and **Levent Kayrin**
Cukurova University, Turkey

Statement of the Problem: β-thalassemia is one of the most monogenic autosomal recessive disorders characterized by defective production of the β-chain of hemoglobin. Definition of the β-globin genotype is necessary for genetic counseling in the carriers, and for predicting prognosis and management options in the patients with thalassemia. DNA-based prenatal diagnosis of β-thalassemia routinely relies on polymerase chain reaction (PCR) and gel electrophoresis. The aim of this study is to develop a new procedure, a DNA-based piezoelectric biosensor, for the detection of β-thalassemia IVSI-110 mutation fetuses cell free DNA from maternal blood, the most common β-thalassemia mutation in Turkey.

Methodology & Theoretical Orientation: Cell-free fetal DNA was taken from maternal whole blood. Bioactive layer was constituted by binding 2-hidroxymetacrilate metacriloamidoscystein (HEMA-MAC) nano-polymers on the electrode's surface. Single oligonucleotide probes specific for IVSI-110 mutation of β-thalassemia were attached to the nano-polymer. The measurements were executed by piezoelectric resonance frequency which is caused by binding of the cell-free fetal DNA in media with single oligonucleotide probe on the electrode surface. The results were confirmed by the conventional molecular method as ARMS.

Findings: The piezoelectric resonance frequencies obtained by hybridization of the cell free fetal DNA on bioactive layer were found to be 216±12, 273±6, and 321±9 Hz for the samples of normal β-globin, heterozygote, and homozygote of IVSI-110 mutation, respectively.

Conclusion & Significance: The developed biosensor serves as a specific result to IVSI- 110 mutation. It could accurately discriminate between normal and IVSI-110 mutation samples. Because of low costs, fast results, specificity and high detection/information effectiveness as compared with conventional prenatal diagnosis methods, we can offer this technique as an alternative to conventional molecular methods.

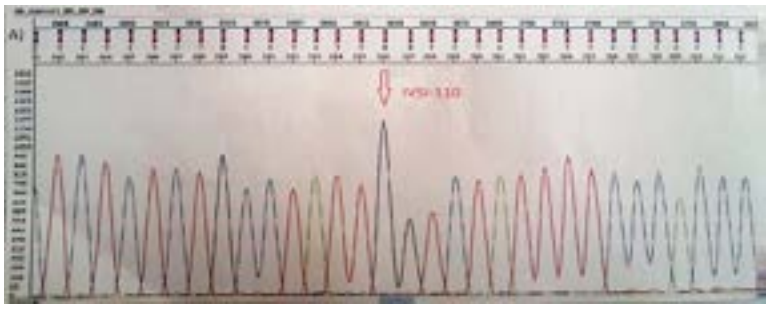

Figure 1: Conventional molecular methods a) sequencing b) gel electrophoresis

Biography

Umut Kokbas has studied Biotechnology and Biochemistry at Ege University. He is a Research Assistant in Medical Biochemistry department at Cukurova University, working on Thalassemia, which is the most common genetic disorder in Turkey. He is also pursuing PhD in the same department.

ukokbas@cu.edu.tr