# Molecular Genetics of β-thalassemia in the Eastern Province of Saudi Arabia

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## Summary

Hemoglobinopathies are the most commonly inherited genetic disorders encountered among the population of the Eastern Province of Saudi Arabia. The thalassemia disease has two major forms known as α and β-thalassemia, and are characterized by quantitative deficiency of either α-globin or β-globin respectively. The high frequency and heterogenicity of β-thalassemia constitutes a major health problem. The objective of this project was to establish the molecular genetics of β-thalassemias in the Eastern Province. Genome DNA was isolated from the blood of 25 patients suspected of being carriers of β-thalassemia. Multiplex PCR for amplification of 4 regions in β-globin gene was undertaken.

The data show that IVS-2 nt 1 and IVS-1 nt 110 mutations are the most common in the population. Codon 39 mutation was also found with relatively high incidence. One of the subjects had a compound mutation which included IVS-2 nt 1 and IVS-1 nt 110 mutations. Six of the subjects with high Hbg $A_2$ showed none of the five common mutations. The results demonstrate the feasibility of using this cost effective technique to screen for the mutation as a pre-marriage requirement.