# Abstract: PB2542

# Title: MOLECULAR SPECTRUM OF ALPHA THALASSEMIA MUTATIONS IN THE WESTERN PROVINCE OF SAUDI ARABIA AND RECOMMENDATION FOR PREMARITAL SCREENING

**Abstract Type: Publication Only**

**Session Title: Thalassemias**

**Background:**

Background: □-thalassemia is frequently encountered in Saudi Arabia . There is a large diversity and geographical variability in underlying genotypes in □-thalassemia patients. HGB H disease can be reduced by premarital thalassemia screening.

**Aims:**

To characterize the molecular spectrum of α -thalassemia in some Saudi patients suspected of α thalassemia carrier or diagnosed with HGB H disease to determine if there is significance of premarital testing for α thalassemia mutations by PCR in couple with suspected α thalassemia trait .

**Methods:**

***Patients and methods*** This study included 39 patients , 34 patients with suspected α thalassemia carrier by having unexplained microcytic hypochromic erythrocytes, with normal iron status and Hb A2 below 3.5%, or being the available parent of a patient with Hb H disease , and 5 patients diagnosed with HGB H disease by HPLC. The 39 patients were screened for 21 α-globin mutations based on polymerase chain reaction (PCR) and reverse-hybridization.

**Results:**

Thirteen patients 33.3% are heterozygous for □ $^{+}$ thalassemia having the genotype -□ $^{3.7}$/ □□ and 14 patients 35.9% are homozygotes for □$^{0}$ having the genotype -□ $^{3.7}$/-□ $^{3.7}$ and, one patient with –$^{MED}$ / □□, one with --$^{SEA}$ / □□, and one – $^{PA-1}$ / □□ and the 5 patients with HGB H disease 12.8% (0.01% from 30200 HPLC results) . one patient has genotype of --$^{SEA}$ /-□ $^{3.7}$ have (3.7 Kb deletion) associated with –$^{SEA}$ . and one with --$^{MED}$/-□ $^{3.7}$ a Mediterranean thalassemia ( --$^{MED}$) with3.7 kb heterozygous deletion, , and 2 patients with genotype α$^{PA-1}$ α/ α$^{PA-1}$α homozygous mutation Poly A (A->G). There was one case with negative molecular screening for □-thalassemia

**Summary/Conclusio**

The -α$^{3.7}$ was the most common mutation among patients with □-thalassemia forms 69.2 % of all deletions . Del. 3.7 kb is not considered as risky mutation . The premarital genetic diagnosis of □-thalassemia is not recommended in Saudi population as the most common α-thalassemia mutation are -α$^{3.7}$ deletion which is not risk for hydrops fetalis . Premarital screening should be considered in families with history of HGB H disease or hydropes fetalis and should include both testing for common deletion and mutation DNA sequencing.

**Keywords:** Thalassemia, Molecular