**Title:** Role of HBS1L-MYB intergenic region polymorphism in thalassemia intermedia patients

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**Abstract:**

**Introduction:** The β-globin disorders are among the most common mendelian disorders in humans. Fetal haemoglobin (HbF) level modifies the clinical severity of HBB disorders. HBS1L-MYB intergenic polymorphism (HMIP) on chromosome 6q23 is associated with elevated fetal haemoglobin levels. Therefore this region is important in the view of presentation of clinical symptoms in β-Thalassemia patients.

**Material and Methods:** In recent study 51 beta Thalassemia Intermediate (TI) patients were evaluated. This particular single nucleotide polymorphism (rs 9399137) that is located on the upstream of MYB gene affects the rate of MYB expression and production of HbF. This SNP was determined by Tetra Primer ARMS-PCR technique and at the end samples were analyzed by agarose gel.

**Results:** To this point in time we have observed bands in the expected length at electrophoresis using designed primers in PCR technique for patient samples. The polymorphism association with TI is currently under investigation.

**Conclusion:** The related HBS1L-MYB polymorphism was observed in TI patients. According to our present observation (not statistically) HBS1L-MYB polymorphism has association with reduction of clinical symptoms.

Thalassemia, HBS1L-MYB intergenic region, fetal haemoglobin, Tetra Primer ARMS-PCR

Presentation: Poster