Title: Study of the association between (C/T) polymorphism in intron 2 of BCL11A gene and HbF level in beta thalassemia intermedia disease

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Abstract: Introduction: Thalassemias are the most common type of hereditary hemoglobinopathies in the world, that are very heterogeneous at the molecular level. Beta thalassemia is caused by point mutations or rarely deletions in beta globin gene cluster, that leads to decreased synthesis (β<sup>+</sup>) or absence (β<sup>0</sup>) of β-globin chains. Increased levels of fetal hemoglobin (HbF) can ameliorate the clinical severity of beta thalassemia intermedia disease. Some polymorphisms in intron 2 of BCL11A gene, a regulator of fetal to adult hemoglobin switching, affect the production and formation of fetal hemoglobin and prevent from clinical symptoms of disease. In the present study, the association between increased levels of HbF and the amelioration of beta thalassemia intermedia disease has been investigated.<br />

Method: Common polymorphism rs11886868 of BCL11A gene was genotyped by Tetra-Primer ARMS PCR method in 50 beta thalassemia intermedia patients. Then PCR products were analysed by agarose gel electrophoresis.<br />

Results: Expected bands were observed and genotyped in samples. Initial observations showed that this polymorphism associated with decreased severity of disease, and it's relationship with HbF expression is under investigation.<br />

Conclusions: Detection of the factors that induce more expression of HbF, is important in beta thalassemia intermedia disease. So if there is a statistical correlation, with initial genotyping of patients, we can informed professionals in prescribing and blood transfusion.<br />

Beta thalassemia intermedia, BCL11A, Polymorphism, HbF

Presentation: Poster