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Identification of Quantitative Trait Loci (QTLs) for Fetal Hemoglobin in Thalassemia

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Description

The genetic makeup of fetal hemoglobin and the modifier genes that control the ratio of alpha to non-alpha globin chains, which is necessary to ameliorate the beta thalassemia phenotype. By doing this, the disease's severe phenotype is avoided. From severe transfusion-dependent thalassemia major to non-transfusion-dependent asymptomatic instances, people with β-thalassemia have presented with a variety of clinical manifestations. It is already well established that patients with TM who had greater Fetal Hemoglobin [HbF] levels also had less severe disease phenotypes [1]. At the moment, secondary and tertiary genetic modifiers that may improve the clinical phenotype of β-thalassemia syndromes are the focus of most research. When it comes to HbF, the most prevalent Quantitative Trait Loci (QTLs) include genes such as: Leukemia/B-cell Lymphoma 11A (BCL11A), Krueppel-Like Factor 1 (KLF1) [2]. Examining the genes and genetic modifiers that control the balance and imbalances of the alpha-beta globin gene clusters is essential to comprehending the phenotypic variability in thalassemia.

Since *BCL11A* is crucial for hemoglobin switching, any deletions in *BCL11A*-binding motifs were thought to be linked to elevated HbF [3]. Together with the *SOX6* gene, which is well known for reconfiguring the β-globin cluster by selectively modulating chromosomal loop formation, *BCL11A* primarily binds to the upstream locus control region (LCR), γ-globin, and the intergenic regions that exist between β-globin and γ-globin genes. This ultimately results in transcriptional silencing of the γ-globin genes. Numerous studies have shown that individuals with Sequence variations in the *HSB1L-MYB* or *BCL11A* genes are associated with less severe beta-thalassemia and greater amounts of fetal hemoglobin [4].

It is crucial to rely on or look into any element that contributes significantly to lowering the alpha/non-alpha globin chain imbalance and may have an ameliorative influence on the disease's clinical picture in order to better understand the clinical or molecular connections [5]. It has been discovered that a few of the documented mutations in the β-globin promoter region are linked to higher γ-chain expression. In essence, these are acknowledged as primary modifiers. These days, the emphasis is more on the additional genetic variations known as secondary modifiers that affect HbF levels but are not connected to the Hb genes. We refer to these modifiers as secondary modifiers; they mainly work directly to modify the illness's established pathophysiology [6,7].

Alterations in genes influencing the α/β globin chain balance, such as α and γ-globin genes, are considered secondary modifiers. Additionally, genes involved in the expression of the γ-globin gene, such as *HBS1MYB* and *BCL11A*, are taken into consideration [8,9]. Recent years have seen notable advancements in the study of secondary genetic modifiers, which have been shown to improve the clinical manifestation of β-thalassemia. Undoubtedly, a rise in fetal hemoglobin (HbF) synthesis during the adult years may lessen the severity of the β-thalassemia phenotype, as γ-globin polypeptide chains fill the role of the functioning β-globin polypeptide chains [10-13]. Therefore, the most popular targets for contemporary therapeutic interventions are γ-globin genes and other secondary modifiers.

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