كلية الطب – جامعة الفيوم
قسم الأطفال

البحث الرابع

XmnI polymorphism in Egyptian patients with β-thalassemia major and its correlation with the

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Abstract

Clinical severity of β-thalassaemia depends on the types of β-gene mutations involved. It can also be influenced by genetic factors like concomittant α-thalassaemia and increased γ-chain production. Several loci are implicated in higher production of HbF. The XmnI restriction site at −158 position of the Gγ-gene is associated with increased expression of the Gγ-globin gene and higher production of HbF. This study aims to determine the frequency of the Gγ XmnI polymorphism in β-thalassaemia patients in Egypt and its correlation to the HBF level and clinical severity of the disease. We investigated the XmnI polymorphism in 100 children with β-thalassaemia major using polymerase chain reaction (PCR-RFLP)-restriction fragment length polymorphism. We found that ninety-four children had XmnI (−/−) genotype (94%) and six children had XmnI (+/−) genotype. On the other hand, the study found that the presence of this polymorphism influences HbF concentration and ameliorate the clinical severity of the disease.