Molecular characterisation and frequency of $^{G}\gamma$ Xmn I polymorphism in Egyptian β-thalassaemia patients and its correlation to the HBF Level and clinical severity of the disease.

Thesis
Submitted for partial fulfillment of the
M. D. degree in
Clinical and Chemical pathology

By
Heba Mostafa Ahmed Mohammed
(M.B.B.Ch, M.Sc.)

Supervised by

Prof.Dr Somaya Mohammad ELgawhary
Professor of Clinical and Chemical Pathology
Faculty of Medicine, Fayoum University

Prof.Dr Mona Mohamed Farid Konsouh
Professor of pediatrics
Faculty of Medicine, Fayoum University

Dr.Mostafa Ahmed Ezzat
Lecturer of Clinical and Chemical Pathology
Faculty of Medicine, Fayoum University

Faculty of Medicine
Fayoum University
2015

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 *Xmn* I 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 *Xmn* I polymorphism in β-thalassaemia patients in Egypt and its corelation to the HBF Level and clinical severity of the disease. we investigated the *Xmn* I 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 Hb F concentration and ameliorate the clinical severity of the disease .

Keywords : β-thalassaemia , Hb F, Gγ *Xmn* I, PCR-RFLP.