AL-QUDS UNIVERSITY
Deanship of Graduate Studies

Molecular Genetics Analysis of *MEFV* Gene Mutations in Familial Mediterranean Fever (FMF) Among Palestinians in the West Bank

By

Rania Abu Seir

رانيا يوسف ابراهيم ابو سير

9811459

Supervisor: Dr. Hisham Darwish

A Thesis Submitted to the Graduate School of Al-Quds University in Partial Fulfillment of the Requirements for the Masters Degree In Biochemistry and Molecular Biology

AL-QUDS UNIVERSITY

2001

ABSTRACT

Familial Mediterranean fever (FMF) is an autosomal recessive hereditary disease characterized by recurrent self-limited attacks of fever accompanied by peritonitis, pleurisy and arthritis with sterile infiltration of polymorphonuclear leukocytes into the serosal and synovial fluids. The most severe complication of FMF is nephrotic amyloidosis which leads to terminal renal failure at an early age. Life-long colchicine treatment is used for alleviation of acute attacks and for prevention of amyloidosis. The disease affects mostly ethnic groups living around the Mediterranean basin including non-Ashkenazi Jews, Armenians, Turks, and Arabs. Recently, the gene linked to FMF (*MEFV*, MEditerranean FeVer) has been mapped to the short arm of chromosomes 16. This gene encodes for the pyrin/marenostrin protein, which is thought to play a role in granulocyte-mediated inflammation.

In this study, *MEFV* gene mutations were investigated in sixty-seven Palestinian patients from different localities of the West Bank that were clinically diagnosed as having FMF. DNA was extracted from EDTA-blood samples and analysis of *MEFV* gene mutations was performed using Polymerase Chain Reaction (PCR), Amplification Refractory Mutation System (ARMS) and direct DNA sequencing.

Twelve missense mutations were identified in 71% of the 134 independent alleles, with both FMF alleles identified in 62.5% of the patients. Eight of these mutations are located in exon ten, two in exon two, and two in exon 3. Evidently, the M694V mutation appears to be the most common one accounting for 31% of all