بسم الله الرحمن الرحيم

MONA MAGHRABY

شبكة المعلومات الجامعية

التوثيق الالكتروني والميكروفيلم

MONA MAGHRABY

شبكة المعلومات الجامعية

التوثيق الإلكتروني والميكروفيلم

جامعة عين شمس

التوثيق الإلكتروني والميكروفيلم

قسم

نقسم بالله العظيم أن المادة التي تم توثيقها وتسجيلها
علي هذه الأقراص المدمجة قد أعدت دون أية تغيرات

يجب أن

تحفظ هذه الأقراص المدمجة بعيدا عن الغبار

MONA MAGHRABY

Thiopurine S- methyltransferase genotype mutation in pediatric patients with inflammatory bowel disease on azathioprine: A pilot study

Thesis

Submitted For partial Fulfillment of

MD Degree in Pediatrics

By

Nada Atef Abd El-Hafez

M.B.B.CH(2008) M.Sc(2013)

Faculty of Medicine - Ain Shams Univeristy

Under Supervision of

Prof. Dr. Mostafa Abd Al-Aziz El-Hodhod

Professor of Pediatrics

Faculty of Medicine - Ain Shams Univeristy

Dr.Marwa Talaat El-Deeb

Assistant Professor of Pediatrics

Faculty of Medicine - Ain Shams Univeristy

Dr.Yosra Mohamed Mohsen

Lecturer of Pediatrics

Faculty of Medicine - Ain shams Univeristy

Dr.Nora Nassef Abd El-Gawad

Molecular Genetics Researcher

Molecular Genetics and Enzymology Dep.

National Research Centre

Faculty of Medicine - Ain Shams Univeristy

2020

بسم الله الرحمن الرحيم

قالوا

سبحانك لا علم لنا إلا ما علمتنا إنك أنت العليم الحكيم

صدق الله العظيم

سورة البقرة الآية: ٣٢

Acknowledgment

First and foremost, I feel always indebted to Allah, the Most Kind and Most Merciful.

I'd like to express my respectful thanks and profound gratitude to ***Prof. Dr. Mostafa Abd El Aziz El Hodhod,*** *Head of Pediatrics department, Faculty of medicine- Ain Shams University for keen guidance, kind supervision, valuable advice and continuous encouragement his which made possible the completion of this work..*

I am also delighted to express my deepest gratitude and thanks to ***Prof. Dr. Marwa Talaat El deeb,*** *Professor of Pediatrics Faculty of medicine- Ain Shams University, for her kind care, continuous supervision, valuable instructions, constant help and great assistance throughout this work.*

I wish to introduce my deep respect and thanks to ***Dr.Yosra Mohamed Mohsen,*** *Lecturer of Pediatrics, Faculty of Medicine, Ain shams Univeristy for her great help, active participation and guidance.*

I wish to introduce my deep respect and thanks to ***Dr.Nora Nassef Abd El-Gawad,*** *Molecular genetics Researcher, Molecular genetics and enzymology Dep., National research center for her kindness and supervision.*

Special thanks to ***Prof. Dr. Ahmad Mohamed Hamdy,*** *head of gastroenterology department, ain shams university, and the whole gastroenterology team work,* ***Dr Dina Abdel Hamid, Dr Heba Amin and Dr Dina Heidar*** *for their support and encouragement..*

Last but not least my sincere thanks and appreciation to all patients participated in this study from GIT clinic, Children Hospital, Ain shams University.

Dedication

I dedicate my dissertation work to the sake of ***Allah,*** *my Creator and my Master,*

My great teacher and messenger, prophet ***Mohammed*** *(May Allah bless and grant him), who taught us the purpose of life*

My family, friends and professors. A special feeling of gratitude to my loving ***parents,*** *whose words of encouragement and push for tenacity ring in my ears.*

My brother ***Mohammed*** *who stands by me when things look bleak. I also dedicate this dissertation to my* ***husband*** *who leads me through the valley of darkness with light of hope and support. I will always appreciate all they have done, I dedicate this work and give special thanks to my best friend* ***Sara****.* ***Kinda****, my wonderful daughter with her secure hugs, All the people in my life who touch my heart, I dedicate this research.*

List of Contents

List of Tables

List of Tables (Cont…)

List of Figures

List of Figures (Cont…)

List of Figures (Cont…)

List of Abbreviations

Abb.	Full term

6-MMP.....: *6-Mercaptopurine*
6MP.....: *6-Mercaptopurine*
ACCA.....: *Antichitobioside carbohydrate IgA.*
ADA.....: *Adalimumab*
ALCA.....: *Anti laminaribioside carbohydrate IgG*
ALT.....: *Alanine transaminase*
AMCA.....: *Anti-synthetic mannoside antibodies.*
ANC.....: *Absolute neutrophilic count*
ANCA.....: *Antineutrophil cytoplasmic antibodies.*
Anti-C.....: *Anti-chitin*
Anti-L.....: *Anti-laminarin*
Anti-OmpC.....: *Antibodies to outer membrane porin C*
APC.....: *Antigen presenting cells.*
ASA.....: *Amino salicylic acid*
ASCA.....: *Anti-Saccharomyces cerevisiae antibodies*
AST.....: *Aspartate transaminase*
ATG.....: *Autology related Gene*
BMI.....: *Body mass index.*
C14orf119.....: *Chromosome 14 Open Reading Frame 119*
CARD.....: *Caspase activation and recruitment domains*
CBC.....: *Complete blood picture*
CCR.....: *Cassette chromosome recombinases*
CD.....: *Crohn's disease*
CDH.....: *Cellobiose dehydrogenase*

List of Abbreviations (Cont…)

Abb.	Full term
CPEB	*Cytoplasmic polyadenylation element binding protein*
CREM	*cAMP responsive element modulator*
CRP	*C-reactive protein*
DAP	*Death-associated protein*
DNA	*Deoxyribonucleic acid*
DNMT3A	*The DNA methyltransferase 3A*
DRP	*Dynamin-related protein*
ECCO	*European Crohn's and Colitis Organization*
ECM	*Extracellular matrix*
ECM1	*Extracellular matrix protein 1*
EDTA	*Ethylenediaminetetraacetic acid*
EEN	*Exclusive Enteral Nutrition*
ERAP	*The endoplasmic reticulum aminopeptidases*
ESPGHAN	*The European Society for Paediatric Gastroenterology Hepatology and Nutrition*
ESR	*Erythrocyte sedimentation rate*
FADS	*Flavin adenine dinucleotide (FAD)-binding proteins*
FC	*Fecal calprotectin.*
FDFT1	*Farnesyl-Diphosphate Farnesyltransferase*
FGCR2A	*Fc fragment of IgG receptor IIa*
FL	*Fecal lactoferrin.*
GCKR	*The glucokinase regulatory protein*
GI	*Gastro intestinal.*
GMPR2	*Guanosine Monophosphate Reductase*