

✓ OK on Disk 59

Study of genetic aspects of severe visual impairment in Egypt

Thesis
Submitted For Partial Fulfillment Of The Degree of M.D.
In Clinical Genetics

616.042
A. A. By **Amira Abdel Mageed Abdel Azeem**
M.Sc.
Under the supervision of



99/4/1

Prof. Dr. Rabah Mohamed Shawky

Professor of Pediatrics and Genetics
Head of Genetics Unit
Faculty of Medicine - Ain Shams University

57354

Prof. Dr. Moones Abdel Latif Ali

Professor of Ophthalmology
Research Institute of Ophthalmology

مكتبة كريمة

Dr. Mohamed Saad Zaghloul

Assistant Professor of Genetics
Pediatric department
Faculty of Medicine - Ain Shams University

Dr. Othman Ali Othman Zico

Assistant Professor of Ophthalmology
Faculty of Medicine - Ain Shams University

Dr. Mahmoud M. Rifaat

Senior Scientist in Genetic Engineering
Research Institute

Faculty of Medicine
Ain Shams University
1999



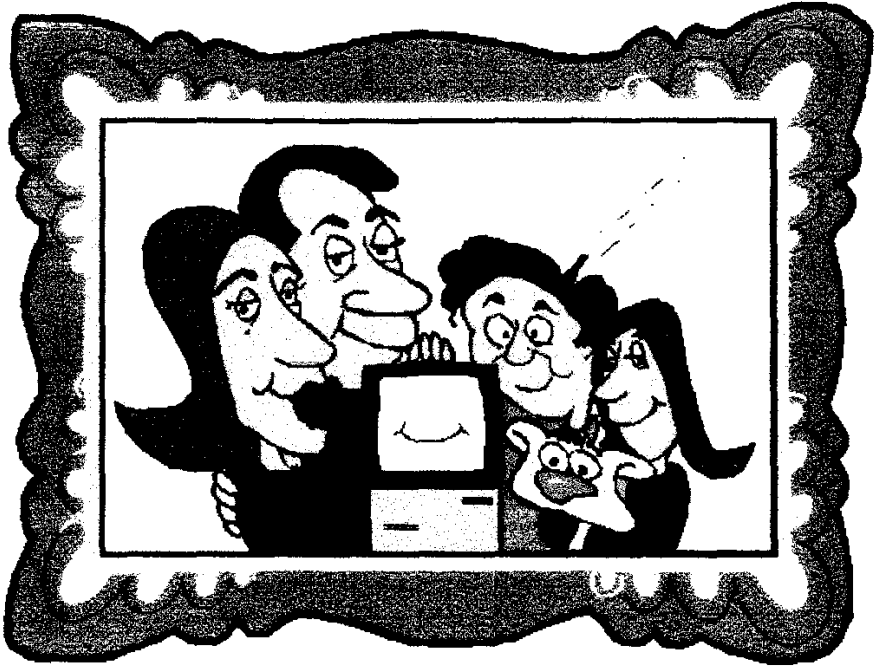


بسم الله الرحمن الرحيم

إنا خلقنا الإنسان من نطفة أمشاج نبتليه
فجعلناه سميعا بصيرا

صدق الله العظيم

سورة الانسان (٢)



TO MY FAMILY



إلى أُمِّي سر و جدوى

وإلى أُمِّي مصرر ثقتي وإيماني

إلى زوجي رمز العطاء

وإلى أخى محمد نبع الوفاء



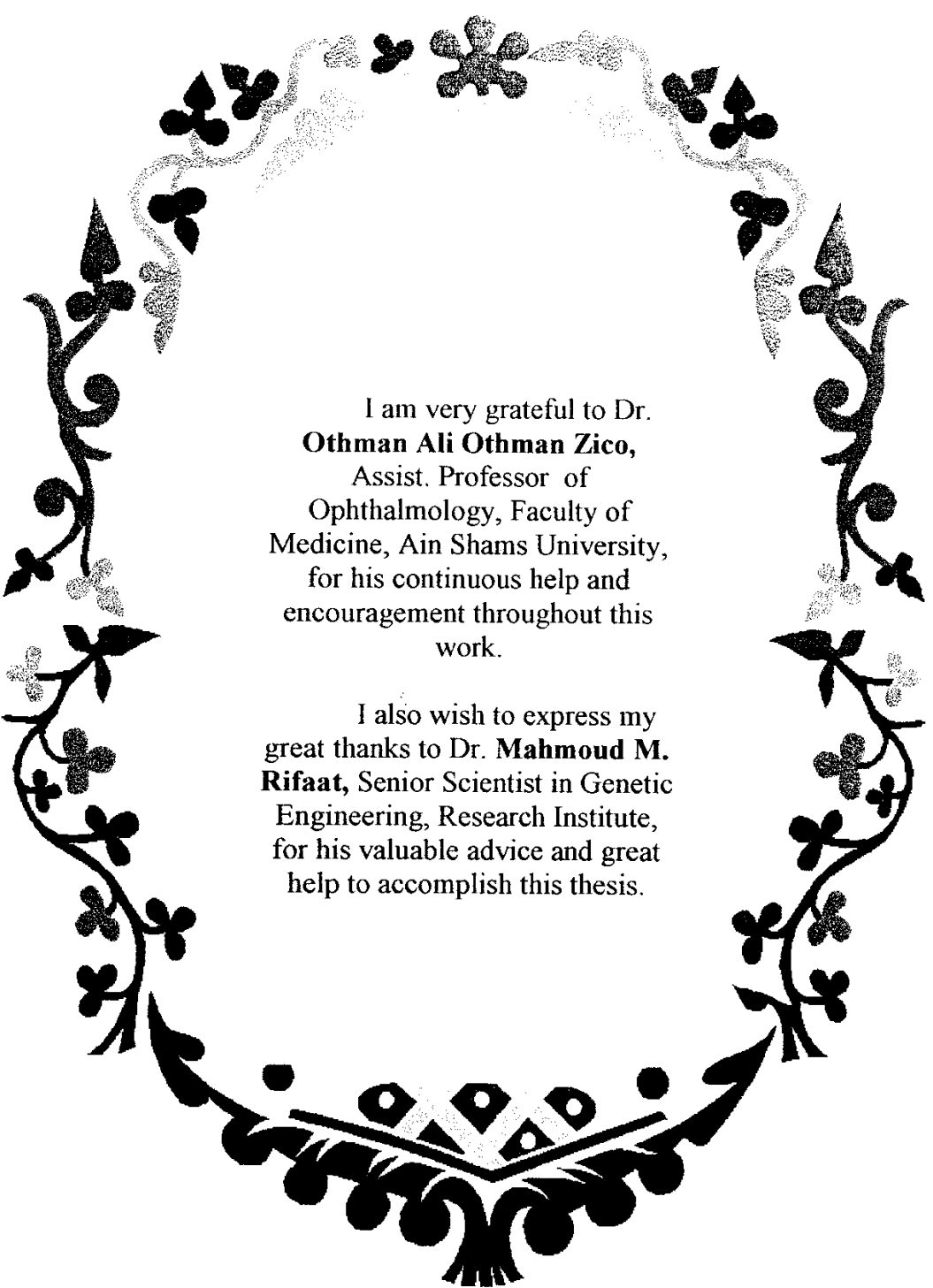


ACKNOWLEDGEMENTS

I would like to express my sincere gratitude to Prof. Dr. **Rabah Mohamed Shawky**, Professor of Pediatrics and Genetics, and Head of Genetics Unit, Faculty of Medicine, Ain Shams University, for her continuous guidance, great help, extreme patience and endless encouragement throughout the whole work.

I am greatly indebted to Prof. Dr. **Moones Abdel Latif Ali**, Professor of Ophthalmology, Research Institute of Ophthalmology, for his great help and ultimate support in preparing this work.

My special great thanks to Dr. **Mohamed Saad Zaghloul**, Assist. Prof. of Genetics, Faculty of Medicine, Ain Shams University, who was very generous in time and effort during the preparation of this work. His great help can never be forgotten.



I am very grateful to Dr.
Othman Ali Othman Zico,
Assist. Professor of
Ophthalmology, Faculty of
Medicine, Ain Shams University,
for his continuous help and
encouragement throughout this
work.

I also wish to express my
great thanks to Dr. **Mahmoud M.
Rifaat**, Senior Scientist in Genetic
Engineering, Research Institute,
for his valuable advice and great
help to accomplish this thesis.

CONTENTS

	Page
- List of Tables	iii
- List of Figures	vii
- List of Abbreviations	xiv
- Introduction and Aim of the Work	1
- Review of Literature	
I- Anatomy and Physiology of Visual System	
1- Structure and function of the eye	3
2- Visual pathway	7
3- Physiology of vision	9
II- Ocular Embryology	13
III- Diagnostic Tests of Vision	
1- Estimation of visual acuity	18
2- Assessment of visual field	26
3- Electrodiagnosis	31
IV- Genetic Diagnostic Techniques	
1- Cytogenetic techniques	34
2- Molecular techniques	39
V- Definition and Etiological Classification of Visual Impairment	
1- Definition	52
2- Etiological classification of visual impairment	54
- Acquired causes	
- Genetically determined causes	

	Page
VI- Genetically Determined Retinal Dystrophies	60
VII- Retinoblastoma	94
VIII- Glaucoma	110
IX- Cataract	124
X- Optic Atrophy	139
XI- Corneal dystrophies	148
XII- Genetically Determined Ocular Malformations	151
- Subjects and Methods	160
- Results	175
- Discussion	249
- Summary	272
- Conclusion	277
- Recommendations	278
- References	279
- Arabic summary	