MOLECULAR STUDIES & GENOTYPE PHENOTYPE CORRELATION IN EGYPTIAN β-THALASSEMIA PATIENTS

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For The Degree In Paediatrics

By

Ghada Youssef El Kammah

MB,BCh. , MSc. (Pediatrics)

Supervised By

Prof. Dr. Samia A Temtamy

Prof. Of Human Genetics

Human Genetics Dept.

National Research Centre

Prof. Dr. Amal El Beshlawy

Prof. Of pediatrics

Faculty Of Medicine

Cairo university

Prof. Dr. Mostafa El Nashar

Prof. Of ENT

Institute of Childhood Studies

Ain Shams University

Prof. Dr. Mostafa El Awady

Prof. Of Biochemical & Molecular Genetics

Head of Human Genetics Dept.

National Research Centre

INSTITUTE OF CHILDHOOD STUDIES

AIN SHAMS UNIVERSITY

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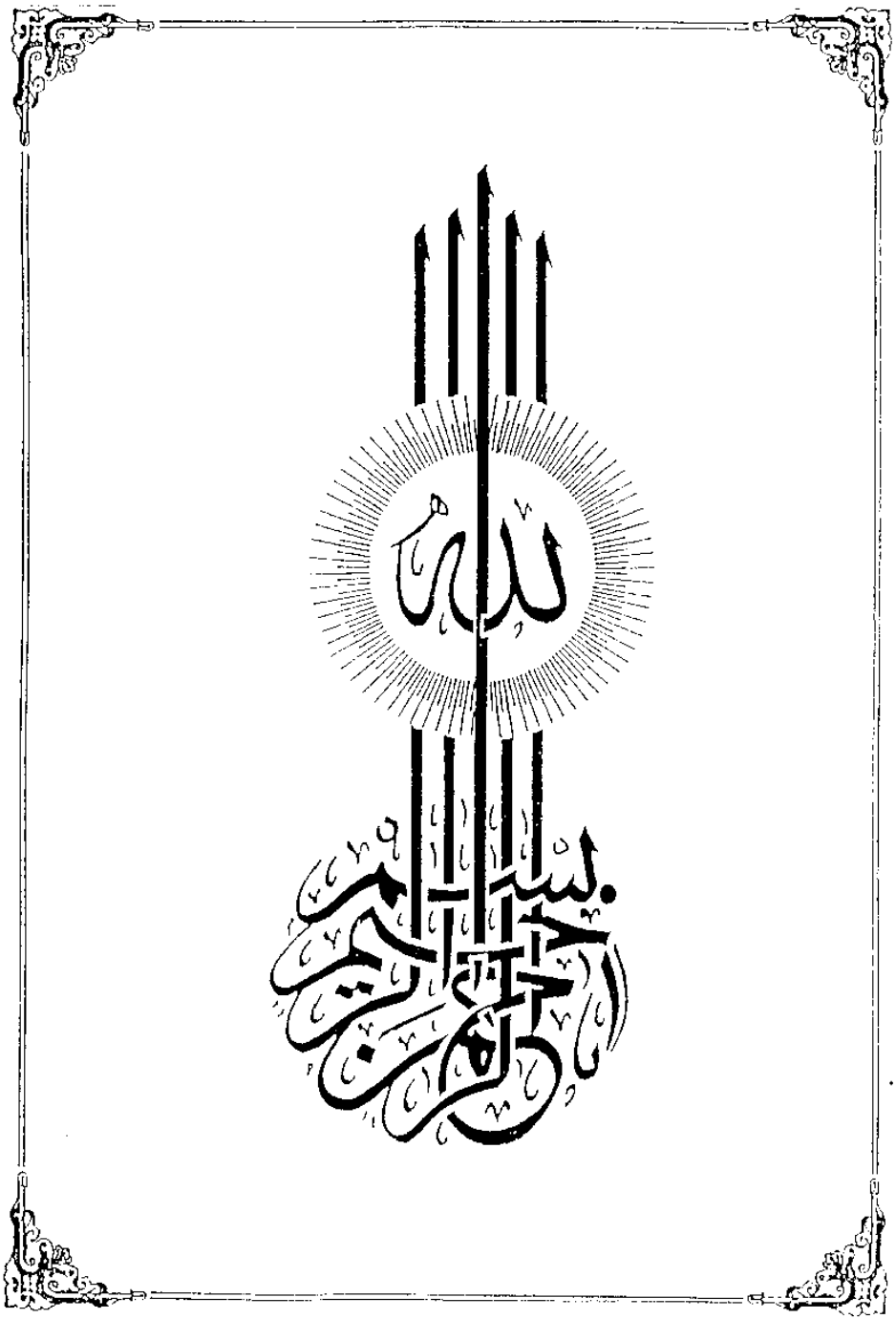

AIN SHAMS UNIVERSITY
INSTITUTE OF CHILDHOOD STUDIES

Name of the candidate : Ghada Youssef El-Kamah

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Discussion and judgment committee

1- Prof. Dr. Samia A Temtamy	Prof. of Human Genetics Human Genetics Dept. Human Genetics Dept.
2-Prof. Dr. Rabah Shawki	Prof. of pediatrics and Human Genetics Faculty of Medicine Ain Shams university
3-Prof. Dr. Mostafa El Nashar	Prof. of ENT Institute of Childhood Studies Ain Shams University
4-Prof. Dr. Lamis Ragab	Prof. of pediatrics Faculty Of Medicine Cairo university

Date of discussion : - / - / 1998
Higher studies and research

Agreement **Agreement**

Approval of councils **Approval of council's university**

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CONTENTS

page

-Introduction and Aim of The Work 1

-Review of Literature

Chapter I : Historical background 3

Chapter II : Normal human hemoglobin 6

-Structure of adult hemoglobin 6

-Structure of the β-globin gene structure 9

*General structural features of globin genes 10

*Control of the β-globin gene expression 12

*Brief recall of the β-globin gene expression 14

-Ontogenic regulation of globin gene 18

Chapter III : Hemoglobinopathies 21

-Structural hemoglobin variants 23

*Variants causing hemolytic anemia 23

*Mutants with altered oxygen transport 23

*Structural mutants that result in thalassemic phenotype 23

-Thalassemias 27

*Clinical classification 27

*Genetic classification 27

-Hereditary persistence of fetal hemoglobin 30

Chapter IV : Hematological and biochemical diagnosis of β-thalassemia 32

-Homozygous β-thalassemia 33

-Heterozygous β-thalassemia 35
-Some of the biochemical analysis in thalassemic patients 37
Chapter V : DNA technology in the diagnosis of β-thalassemia 38
Chapter VI : Molecular pathology of β-thalassemia . . 43
-β-thalassemia gene deletion 49
-Transcriptional mutations 51
-RNA splice site mutations 51
*Involving intron / exon boundaries (splice junction changes) 51
*Splice site consensus sequence mutations 52
*Mutations in cryptic sites in exons (coding region) , substitution affecting processing 52
*Mutations at cryptic in introns 53
-Cap site mutations 54
-Polyadenylation signal mutation 54
-Mutations resulting in abnormal translation of the β-globin mRNA 55
*mutants producing a termination codon (nonsense mutations) 55
*Frameshift mutations 55
*The third type 56
-Unstable β-chain variants 56
-Silent β-thalassemia 57
-Deletion mutations in β-locus control region (εγδβ)° thalassemia 57

-Deletions causing δβ-thalassemia or HPFH 57
-Thalassemia like disorders due to two mutations in the β-globin gene cluster 58
*Hemoglobin with novel physical properties : Hb C . 59
Chapter VII : Clinical aspects of β-thalassemia 60
-Thalassemia major 60
-Thalassemia intermedia 64
-Heterozygous β-thalassemia 66
-Dominantly inherited β-thalassemia 66
Chapter VIII: Genotype / phenotype correlation in β-thalassemia patients 67
-Effect of gross deletions in the β-globin gene cluster . 68
*Hereditary persistence of fetal hemoglobin 68
*Heterozygous δβ-thalassemia 68
*γδβ-thalassemia 68
-Phenotype of heterozygous β-thalassemia 69
*Heterozygous β-thalassemia with normal red cell indices 69
*Heterozygous β-thalassemia with normal Hb A2 . . 70
*Silent β-thalassemia 71
*Heterozygous β-thalassemia with unusually high Hb A2 Level 72
*β-globin chain variants coinhrited with heterozygous β-thalassemia 72
-Homozygous β-thalassemia 72
*Phenotype of mild β-thalassemia mutations . . . 72

*phenotype of homozygous β-thalassemia coinherited with α-thalassemia 76
*Phenotype of β-thalassemia homozygous with enhanced γ-chain production 76
Chapter IX : The control and treatment of β-thalassemia .79
-Carrier screening 79
-Genetic counseling 80
-Prenatal diagnosis 80
-Management of β-thalassemia cases 81
*Transfusion therapy 81
*Iron chelation therapy 82
*Splenectomy 83
*Bone marrow transplantation (BMT) 84
*Experimental procedures 85
-Patients and Methods 88
-Results 92
-Discussion 126
-Conclusion and Recommendations 146
-Summary 148
-References 151
-Arabic Summary . . . . . .

List of Abbreviations

a.a.	: Amino acid .
A.	: Adenin .
Aγ	: Gumma chain with alanine at position 136 of the polypeptide chain .
ARMS	: Amplification refractory mutation system .
ASO	: Allele specific oligonucleotides .
BMT	: Bone marrow transplantation .
BSU36	: Type of a restriction enzyme .
bp	: Base pair .
C	: Cytosine .
CAP	: Catabolite activator protein .
CHABA	: Congenital Heinz bodies hemolytic anemia .
CVS	: Chorionic villus sampling .
DGGE	: Denaturing gradient gel electrophoresis .
DNA	: Deoxyribonucleic acid .
Dnase	: Deoxyribonuclease .
EcoRI	: Type of restriction enzyme .
EPO	: Erythropoietin .
G	: Guanin .
Gγ	: Gumma chain with glycin at position 136 of the polypeptide chain .
GVHD	: Graft versus host disease .
HMZ	: Homozygous .
HPFH	: Hereditary persistence of fetal hemoglobin .
HS	: Hypersensitive site .

HSC	: Hematopoietic stem cells .
HTZ	: Heterozygous .
IVS	: Intervening sequence .
Kb	: Kilo base .
LCR	: Locus control region .
MCH	: Mean corpuscular hemoglobin .
MCV	: Mean corpuscular volume .
mRNA	: Messenger ribonucleic acid .
nts	: Nucleotides .
PCR	: Polymerase chain reaction .
PTH	: Parathyroid .
RBC	: Red blood corpuscle .
RDW	: Red cell distribution width .
RE	: Restriction enzyme .
RES	: Reticulo endothelial system .
RFLP	: Restriction fragment length polymorphism .
RNP	: Ribonucleoprotein .
RPI	: Reticulocyte production index .
SSCP	: Single stranded conformation polymorphism .
T	: Thymidine .
T3	: Tri-iodo thyronine .
T4	: Thyroxine .
tRNA	: Transfer ribonucleic acid .
TSH	Thyroid stimulating hormone .
U	: Uracil
Unch.	: Uncharacterized .

List of Figures

Fig		page
1	: Schematic diagram of quaternary structure of hemoglobin and the relative changes in orientation of the subunits during oxygenation.	8
2	: Alpha and beta globin gene cluster	11
3	: Genetic control of human hemoglobin	17
4	: Globin synthesis at various stages of embryonic and fetal development	19
5	: Point mutations in the β-globin gene	48
6	: Deletions affecting the β-globin gene cluster	50
7	: Pathophysiology of β-thalassemia	63
8	: Mild β-thalassemia mutations	74
9	: Correlation between age of onset and frequency of blood transfusion	118
10	: Frequency of β-thalassemia alleles	119
11	: Consanguinity in the most common mutations	120
12	: Height in the most common mutations	121
13	: Digestion by BSU 36 enzyme	122
14	: Example of PCR products from 3 PCR reactions	123
15	: Examples of RDB results (A)	124
	(B)	125

LIST OF TABLES

Table		Page
1	: Genetic disorders of hemoglobin	22
2	: Molecular mechanisms underlying globin mutations	24
3	: The major classes of hemoglobin structural variants	25
4	: The thalassemias and related disorders	28
5	: Inheritance risk for a hemoglobin variant	31
6	: Phenotypic characteristics of β-thalassemia	36
7	: Molecular pathology of β-thalassemias	44
8	: β-thalassemia intermedia	75
9	: Sequence of PCR primers	91
10	: Clinical and hematological criteria of cases studied	98
11	: Frequency of β-thalassemia alleles in the studied cases	102
12	: Genotype frequencies among the studied cases	103
13	: Number and percentage of males and females in common mutations	104
14	: Number and percentage of positive and negative consanguinity in common mutations	105
15	: Range of splenomegaly and number and percentage of cases which had undergone splenectomy	106
16	: Standard deviation of height in cases of common mutations	107
17	: Clinical and hematological data of some rare mutations	108
18	: Clinical and hematological data of four patients homozygous for IVS2 nt 745 and frameshift 5 mutations	109
19	: Comparison between the mean values and standard deviation of the clinical and hematological parameters of	