بسم الله الرحمن الرحيم

شبكة المعلومات الجامعية

جامعة عين شمس

التوثيق الالكتروني والميكروفيلم

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﴿ سُبحَانكَ لَا عِلمَ لَنَا إِلَّا مَا عَلَّمتَنَا
إِنَّكَ أَنتَ العَلِيمُ الحَكِيمُ ﴾

صدق الله العظيم

"سورة البقرة: الآية ٣٢"

GENETIC SCREENING FOR SICKLE CELL DISEASE AND GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY IN NEWBORN INFANTS

A Thesis

Submitted to Medical Research Institute

Alexandria University

In Partial Fulfillment for the Requirements of

PH.D. Degree in Genetics

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Acknowledgement

My deepest gratitude and great appreciation to ***Prof. Dr. Suzan Roushdy Ismail****, professor of human Genetics, Human Genetics Department, Medical Research Institute, Alexandria University, for her unlimited and talented efforts to accomplish this work. She gave me so much of her precious time. Without her helpful guidance, kind encouragement, and constructive comments, this work simply could not be completed.*

I am greatly indebted to ***Prof. Dr. Mervat Moustafa Hashish****, professor of Human Genetics, Human Genetics Department, Medical Research Institute, Alexandria University, and I would like to express my extreme gratitude, and sincere appreciation for her excellent supervision, useful advice, and valuable cooperation all over the course of the work. .*

My deep thanks and gratitude are especially expressed to Dr. ***Mohamed Mohamed Mokhtar****, Assistant Prof. of Human Genetics, Human Genetics Department, Medical Research Institute, Alexandria University, for his continuous advice, helpful follow up, and valuable assistance throughout the course of the work.*

I would like to sincerely acknowledge and thank Dr. ***Homam Mohamed Sharshira****, Assistant Prof. of Hematology, Department of Hematology, Medical Research Institute, Alexandria University, for helping and guiding me during performance of the practical work, and kindly providing the results of the quantitative assay of G-6-PD.*

ABBREVIATIONS

2,3-DPG	2,3-Diphosphoglycerate
G6PD	Glucose-6-phosphate dehydrogenase
GSH	Reduced glutathione
GSSG	Oxidized glutathione
Hb	Hemoglobin
HbA	Normal adult hemoglobin
HbF	Fetal hemoglobin
HbS	Sickle hemoglobin
HPFH	Hereditary persistence of fetal hemoglobin
ICSH	International Council for Standardization in Hematology
LCR	Locus control region
NADP	Nicotinamide-adenine dinucleotide phosphate
NADPH	Reduced nicotinamide-adenine dinucleotide phosphate
PCR	Polymerase chain reaction
PKU	Phenylketonuria

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INTRODUCTION

INTRODUCTION

The hereditary anemias present a major genetic health problem that contributes considerably to childhood mortality and morbidity in many developing countries. They are among the commonest of the genetically determined diseases and comprise a group of conditions of considerable complexity. However, because of the easy accessibility of the red blood cells, more has been learnt about the genetic and molecular basis of anemias than about other inherited human disease. Many of hereditary anemias are rare and are not important as regards public health. However, two groups, the inherited disorders of hemoglobin (hemoglobinopathies) and deficiency of the red cell enzyme glucose-6-phosphate dehydrogenase (G6PD), have achieved an extraordinarily high frequency in the world populations.[1]

The hemoglobinopathies and G6PD deficiency are the most common single gene disorders encountered in our region; they are much more common than in many other parts of the world. They represent a major health problem and the chronic illness and complications of these conditions pose considerable burdens on our health resources. [2]

Sickle cell anemia is a chronic and often debilitating disease, which results from homozygosity for a point mutation in the β-globin subunit of the hemoglobin molecule. Hemoglobin S (HbS), the product of this mutation, polymerizes when deoxygenated, thus damaging the red blood cell and causing vaso-occlusive