GENETIC SCREENING FOR COMMON ALPHA THALASSEMIA MUTATIONS IN CORD BLOOD SAMPLES AMONG EGYPTIAN NEWBORNS

Thesis
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By

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Dedication

TO MY FAMILY

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Abbreviation list

2,3-BPG 2,3-bisphosphoglycerate

Arg Argenine
ASN Antisense
Asp Aspartic acid

ATMDS Alpha thalassemia with myelodysplastic syndrome
ATR-16 Alpha thalassemia mental retardation 16 syndrome

ATRX Alpha thalassemia x linked mental retardation syndrome

Bp Base pair

CBC Complete blood count CMC Carboxymethyl cellulose

CS Constant Spring

CTR Cardio thoracic ration

Cys Cysteine

DC Dissociation curve

DHPLC Denaturation High performance liquid chromatography

DNA Deoxyribonucleic acid

ECFCS Erythroid colony forming cells

Fil Filipino

FOG1 A type of transcription factors
GATA A family of transcription factors
GATA1 A type of transcription factors

Gln Glutamine
Glu Glutamic acid

Gly Glycine

Hb Hemoglobin

HbA Adult hemoglobin
HbF Fetal hemoglobin

Hct Hematocrit (packed red cell volume)

His Histidine

Hph 5 bp deletion involving the 5' splice junction of IVS1

HPLC High performance liquid chromatography

IDA Iron deficiency anemia

Ig Immunoglobulin
IM Intramuscular
IV Intravenous

IVS Intervening sequence

Kb Kilobase

Kbp Kilo base pair

LCR Locus control region

Leu Leucine Lys Lysine

MCH Mean corpuscular hemoglobin

MCHC Mean corpuscular hemoglobin concentration

MCV Mean corpuscular volume MDS Myelodysplastic syndrome

MED Mediterranean

PA-1 Poly A1

PCR Polymerase chain reaction

Pro Proline PS Paksé

QF-PCR Quantitative fluorescence polymerase chain reaction

QS Quong Sze

RBC Red blood cells

RDW Red cell distribution width

RFLP Restriction fragment length polymorphism

RNA Ribonucleic acid

RP-HPLC Reverse phase high performance liquid chromatography

SEA South Eeast Asia

Ser Serine SN Sense

SSM Slipped strand mispairing

STR Short tandem repeat
Ter Termination codon

Thai Thailand

THAL Thalassemia

Tyr Tyrosine

UAE United arab emirates
UTR Untranslated region

Val Valine

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