SYSTEMATIC DIAGNOSTIC APPROACH TO A SPECTRUM OF RED CELL ABNORMALITIES IN UNDIAGNOSED ANEMIAS IN EGYPTIAN CHILDREN AND ADOLESCENTS

Thesis

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List of Abbreviations

Abb.	Full term

AA.....*Aplastic anemia*
ACP..... *Aceruloplasminemia*
ADA.....*Adenosine deaminase*
BTT.....*Beta thalassemia major*
cb.....*Cobolamin*
CDA.....*Congenital dyserythropoietic anemia*
CHC.....*Cryohydrocytosis*
CP.....*Ceruloplasmin*
CRP .....*C reactive protein*
DC.....*Dyskeratosis congenita*
DEB.....*Diepoxybutane*
DIDMOAD.....*Diabetes inspidus, Diabetes Mellitus, Optic atrophy, Deafness*
DM.....*Diabetes Mellitus*
DNA.....*Deoxyribonucleic acid*
EDTA.....*Ethylenediaminetetraacetic acid*
EMA.....*Eosin-5'-maleimide Binding test*
EMS.....*Extended metabolic screen*
FA .....*Fanconi Anemia*
FL.....*Femtoliter*
FP.....*Familial pseudohyperkalemia*
G6PD.....*Glucose-6-phosphate dehydrogenase*
GPI.....*Glucose-6-phosphate isomerase deficiency*
GR.....*Glutathione reductase*
HbA.....*Adult hemoglobin*
HbA2.....*Hemoglobin A2*
HE.....*Herediatary elliptocytosis*
Hgb.....*Hemoglobin*

List of Abbreviations (Cont...)

Abb.	Full term
HIF-1á	*Hypoxia-Inducible Factor 1*
HNSHA	*Hereditary nonspherocytic hemolytic anemia*
HPFH	*Hereditary Persistence of fetal hemoglobin*
HPLC	*High performance liquid chromatography*
HPP	*Hereditary pyropoikilocytosis*
HRE	*Hypoxia Responsive Element*
HS	*Hereditary spherocytosis*
HSt:	*Hereditary stomatocytosis*
IRIDA	*Iron refractory iron deficiency anemia*
IVS	*intervening sequence*
LDH	*Lactate dehydrogenase*
Mcg	*Microgram*
MCH	*Mean corpuscular hemoglobin*
MCHC	*Mean corpuscular hemoglobin concentration*
MCV	*Mean corpuscular volume*
MDR1	*Multidrug resistance protein 1*
MDS	*Myelodysplasia*
MMA	*Methyl Malonic Acidemia*
MMC	*Mitomycin C*
MRI	*Magnetic resonance imaging*
MT-2	*Matriptase-2*
NADP	*Nicotinamide adenine dinucleotide*
NGS	*Next generation sequencing*
OS	*Osmotic fragility*
PBL	*Peripheral blood lymphocyte*
PCR	*polymerase chain reaction*
PFK	*Phosphofructokinase*
PKD	*Pyruvate kinase deficiency*

List of Abbreviations (Cont...)

Abb.	Full term
PS	*Phosphatidylserine*
RBC	*Red blood cell*
RCC	*Refractory cytopenia of childhood*
RDW	*Red cell distribution width*
RFT	*Reduced-folate transporter*
RNA	*Ribonucleic acid.*
RS	*Ring sideroblast*
S/β-thalassemia	*Sickle beta thalassemia*
SCA	*Sickle cell anemia*
SCD	*Sickle cell disease*
SD	*Standard deviation*
SDS-PAGE	*Sodium dodecyl sulfate–polyacrylamide gel electrophoresis*
TC	*Transcobolamin*
TF	*Transferrin*
TIBC	*Total iron binding capacity*
WHO	*World Health Organization*

INTRODUCTION

Anemia is defined as hemoglobin (Hgb) concentration and /or red blood cell (RBC) mass less than the 5th percentile for age ***(Oski et al., 2003).***

Table (1): Variations in Hemoglobin Level and Mean Red Blood Cell Volume ***(Matthew and Jason, 2013)***

Age	Hemoglobin level in G per DL (GM per DL)		Mean corpuscular volume in μm^3 (FL)	
	Mean*	Diagnostic of anemia	Mean	Diagnostic of microcytosis
3 to 6 months	11.5 (115)	9.5 (95)	91(91)	74 (74)
6 months to 2 years	12.0 (120)	10.5 (105)	78 (78)	70 (70)
2 to 6 years	12.5 (125)	11.5 (115)	81 (81)	75 (75)
6 to 12 years	13.5 (135)	11.5 (115)	86 (86)	77 (77)
12 to 18 years (female)	14.0 (140)	12.0 (120)	90 (90)	78 (78)
12 to 18 years (male)	14.5 (145)	13.0 (130)	88 (88)	78

Epidemiology of anemia:

Anemia affects 1.62 billion people all over the world,which corresponds to 24.8% of the population. The preschool-age children has the highest percentage, and the lowest percentage is in men **(*De Benoist et al., 2008).***

According to the World Health Organization (WHO), half of the anemia cases diagnosed are due to iron deficiency

(WHO, 2001). However, the prevalence of iron deficiency is 2.5 times much more than that of anemia as anemia is a late indicator of iron deficiency ***(Zimmermann and Hurrell, 2007).***

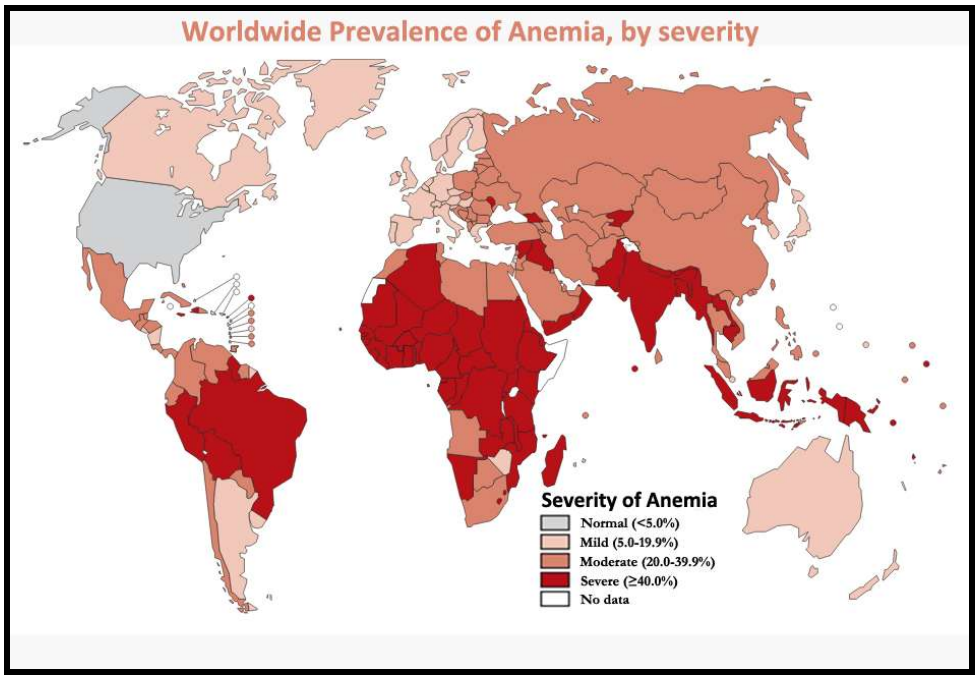

Figure (1): World wide prevalence of anemia by severity ***(WHO, 2015)***

By getting data from the Egyptian Demographic and Health Survey, it was found that between the duration of 2000 to 2005, the prevalence of anemia increased from 37.04% to over 52% between the Egyptian children ranging from 12 months to 36 months of age ***(Austin et al., 2012).*** However, in another study done by Mansour and his colleagues in Quena revealed 12% anemia among school aged children and this may be to iron fortification program ***(Mansour et al., 2004).***