

بِسْمِ اللَّهِ الرَّحْمَنِ الرَّحِيمِ

قال الله تعالى

يَرْفَعِ اللَّهُ الَّذِينَ آمَنُوا مِنْكُمْ وَالَّذِينَ أُوتُوا الْعِلْمَ دَرَجَاتٍ {

صدق الله العظيم

سورة المجادلة الآية 11

## ***Dedication***

*This work is dedicated to those  
who are suffering of Beta-  
thalassemia in Sudan and all  
over the world.*

## *Acknowledgment*

First off all I would like to deep thank Allah for guide me to make this research.

I would like to express my thanks and gratitude to my supervisor

*Dr. Abd El Salam Ibrahim Basheer*, Assistant professor, Department of pathology, University of Khartoum for his never fading support.

I gratefully acknowledge the support of the staff and colleagues at the faculty of medical laboratory sciences, University of Sudan. My appreciation is also offered to *Dr.*

*Sana Eltahir*, Research Centre, University of Aleelain for support and patience. Sincere thanks are extended to *Dr. Mojahid M. Alhassan* and *Dr. Miska Alyman*, Research lab,

University of Sudan for their advices and valued assistance. Deep thanks to my colleague *Mr. Ahmed Fath*

*Elrahman Edris*, Institute of Endemic Diseases, University of Khartoum for his help and support. My thanks extend to members of the Thlassemia Research Group. Above all my special thanks to my family.

## *Abstract*

This a cross sectional study was undertaken at Mabroka village in the Gazeera state between November 2008-February 2009. The study aimed at assessing of the complete blood count and the fetal haemoglobin level in the parents and siblings of beta thalassemia patients. Fetal haemoglobin level was measured by an alkali denaturation procedure. A total number of 50 relatives of beta thlassemia patients were consecutively recruited. The age of the study group were found to be ranged from 10 to 98 year old. The mean of the total haemoglobin level within males was  $12.6 \pm 1.6$  g/dl and within females was  $11.7 \pm 1.4$  g/dl. The mean of the MCV for all population was  $79.6 \pm 8.7$  fl, the mean of the MCH was  $23.8 \pm 3.8$  pg, the mean of the MCHC was  $30 \pm 1.4$  g/dl and the mean of the fetal haemoglobin was  $3 \pm 1.4$  %. The study revealed that, there was no correlation between the age and fetal haemoglobin level among the study population. The results strongly suggested that these participants might be carriers to B-Thalassemia gene.

The study recommended for comprehensive programs in the future to know the actual prevalence of thalassemia in the study population also health education is needed to increase the awareness among that population.

هذه الدراسة المقطعية تم إجرائها في منطقة مبروكة بولاية الجزيرة في الفترة ما بين نوفمبر 2008 إلى فبراير 2009. تهدف الدراسة إلى تقييم مستوى خضاب الدم الجنيني في عينات دم الأشخاص المشاركين فيها. وقد استخدمت طريقة التمسح القلوي لقياس خضاب الدم الجنيني. ويبلغ عدد المشاركين 50 شخص من أسر الأطفال المصابين بأنيميا البحر الأبيض المتوسط البائية. تراوحت أعمار المشاركين في الدراسة ما بين 10 - 98 سنة. بلغ متوسط مستوى خضاب الدم الكلي ومتوسط الهيموغلوبين الجنيني عند الذكور  $12.6 \pm 1.6$  جرام/دسي لتر وعند الإناث  $11.7 \pm 1.4$  جرام/دسي لتر. بلغ متوسط حجم كريات الدم الحمراء  $79.6 \pm 8.7$  فمتو لتر ومتوسط خضاب الدم بالخلية الحمراء  $23.8 \pm 3.8$  بيكوجرام ومتوسط تركيز خضاب الدم بالخلية الحمراء  $30 \pm 3.8$  % جرام/دسي لتر ومتوسط مستوى خضاب الدم الجنيني  $3 \pm 1.4$ .

وكشفت الدراسة عن أنه لا توجد علاقة بين مستوى الهيموكلوبين الجنيني واختلاف العمر. توقت الدراسة بأن الأشخاص المشاركين قد يكونون حاملين لجين مرض الثلاسيميا البائية، ولذا توصي الدراسة بإجراء برنامج مسح مكثف في المستقبل لمعرفة الانتشار الفعلي للمرض في عينة الدراسة بالمنطقة وأيضاً التثقيف الصحي المطلوب لزيادة الوعي بين أولئك الأشخاص.

## ***List of Consents***

إيه قرانية	I
Dedication	II
Acknowledgment	III
Abstract (English)	IV
Abstract (Arabic)	V
List of contents	VI-VII
List of abbreviation	VIII
List of tables	IX
List of figures	X
<b>Chapter One</b> <b>Introduction and literature review</b>	
1.0 Introduction	1
1.1 Haemoglobin	1
1.1.1 Types of normal Haemoglobin	2
1.2 Causes of anaemia	3
1.3 Classification of anaemia	4
1.4 Thalassemias	6
1.4.1 Clinical feature of $\beta$ - thalassaemia	8
1.4.2 Complications of Thalassemias	8
1.4.3 Pathophysiology of $\alpha$ –Thalassemia	9
1.4.4 Epidemiology of $\alpha$ –Thalassemia	10
1.4.5 Pathophysiology of $\beta$ –Thalassemia	11
1.4.6 Epidemiology of $\beta$ –Thalassemia	12
1.4.7 Differential diagnosis of $\alpha$ - & $\beta$ - Thalassemia	12
1.4.7.1 Haemoglobin A2 as diagnostic for $\beta$ -Thalassaemia trait	13
1.4.8 Prenatal Diagnosis of $\alpha$ - & $\beta$ - Thalassemia	14
1.4.9 Treatment of Thalassemia	15
1.4.10 Thalassemia Variants and Related Conditions	16
1.5 Published about thalassemia in Sudan	17
1.6 Hereditary Persistence of Fetal Hemoglobin	17
<b>Chapter Two</b> <b>Objectives and Rationale</b>	
2.0 Rationale	18
2.1 Objectives	19
<b>Chapter Three</b> <b>Material and Methods</b>	
3.0 Materials and methodology	20
3.1 Study design	20
3.2 Study area	20
3.3 Study population	20
3.4 Sample size	20
3.5 Inclusion criteria	20
3.6 Exclusion criteria	20
3.7 Plan of data collection	20
3.7.1 Data analysis	20
3.8 Ethical consideration	21
3.9 Laboratory procedures	21
3.9.1 Materials	21
3.9.1.1 Equipments	21
3.9.1.2 Reagents	22



### *List of abbreviation*

<b>g/l</b>	Gram per liter
<b>g/dL</b>	Gram per deci liter
<b>CBC</b>	Complete blood count
<b>PCV</b>	Packed cell volume
<b>MCV</b>	Mean cell volume
<b>MCH</b>	Mean cell haemoglobin
<b>MCHC</b>	Mean cell haemoglobin concentration
<b>D.W</b>	Distilled Water
<b>OD</b>	Optical density
<b>WHO</b>	World health organization
<b>SD</b>	Standard deviation
<b>Hb</b>	Haemoglobin
<b>Hb F</b>	Fetal Haemoglobin
<b>Min</b>	Minute
<b>Sec</b>	Second
<b>nm</b>	Nanometer
<b>RBCs</b>	Red blood cells
<b>WBCs</b>	White blood cells
<b>Plts</b>	Platelets
<b>2-3 DPG</b>	2,3-diphosphoglycerate
<b>ELISA</b>	Enzyme-linked immunesorbent assay
<b>–ve</b>	Negative
<b>ζ</b>	Zeta
<b>ε</b>	Epsilon
<b>α</b>	Alpha
<b>γ</b>	Gamma
<b>β</b>	Beta
<b>δ</b>	Delta

***List of tables:***

<b>Page No</b>	<b>Table No</b>	<b>Name of table</b>
30	<b>Table 4.1</b>	The distribution of male and female in the study population
31	<b>Table 4.2</b>	The distribution of age groups in the study population
32	<b>Table 4.3</b>	The correlation between the fetal Hb and the age in the population
33	<b>Table 4.4</b>	The haemoglobin and PCV levels according to gender
34	<b>Table 4.5</b>	The MCV, MCH, MCHC and Hb F in the study population
36	<b>Table 4.7</b>	The normal and abnormal total Hb & PCV according to gender
37	<b>Table 4.8</b>	The normal and abnormal MCV within males and females
38	<b>Table 4.9</b>	The normal and abnormal MCH within males and females
39	<b>Table 4.10</b>	The normal and abnormal MCHC within males and females
40	<b>Table 4.11</b>	The means, standard deviation and P. value for fetal haemoglobin according to gender

### *List of figures:*

Page No	Figure No	Name of figure
3	<b>Figure 1.1</b>	Embryonic, Fetal and Adult haemoglobin structure.
3	<b>Figure 1.2</b>	Chromosome 11 & 16 structure.
7	<b>Figure 1.3</b>	Global distribution of haemoglobin disorders.
14	<b>Figure 1.4</b>	The belt of B-Thalassemia.
17	<b>Figure 1.5</b>	Differential diagnosis of Thalassemias.
30	<b>Figure 4.1</b>	The distribution of male and female in the study population.
31	<b>Figure 4.2</b>	The distribution of age groups in the study population.
32	<b>Figure 4.3</b>	The correlation between the fetal Hb and the age in the population
33	<b>Figure 4.4</b>	The haemoglobin and PCV levels according to gender.
34	<b>Figure 4.5</b>	The MCV, MCH, MCHC and Hb F in the study population.
35	<b>Figure 4.6</b>	The degrees of RBCs size in peripheral blood smear of the population.
36	<b>Figure 4.7</b>	The normal and abnormal of total Hb & PCV according to gender.
37	<b>Figure 4.8</b>	The normal and abnormal MCV within males and females.
38	<b>Figure 4.9</b>	The normal and abnormal MCH within males and females.
39	<b>Figure 4.10</b>	The normal and abnormal MCHC within males and females.
40	<b>Figure 4.11</b>	The means, standard deviation and P. value for fetal haemoglobin according to gender.