# بِسْمِ اللَّهِ الرَّحْمَنِ الرَّحِيمِ

### قال الله تعالى

يَا أَيُّهَا النَّاسُ إِنْ كُنْتُمْ فِي رَيْبٍ مِنَ الْبَعْثِ فَإِنَّا خَلَقْنَاكُمْ مِنْ تُرَابٍ
ثُمَّ مِنْ نُطْفَةٍ ثُمَّ مِنْ عَلْقَةٍ ثُمَّ مِنْ مُضْغَةٍ مُخَلَّقَةٍ وَغَيْرٍ مُخَلَّقَةٍ
لِنُبَيِّنَ لَكُمْ وَنُقِرُّ فِي الْأَرْحَامِ مَا نَشَاءُ إِلَى أَجَلٍ مُسَمَّى ثُمَّ نُخْرِجُكُمْ
لِنُبَيِّنَ لَكُمْ وَنُقِرُ فِي الْأَرْحَامِ مَا نَشَاءُ إِلَى أَجَلٍ مُسَمَّى ثُمَّ يُحَرُّ إِلَى
طِفْلًا ثُمَّ لِتَبْلُغُوا اشُدَّكُمْ وَمِنْكُمْ مَنْ يُتَوَقَّى وَمِنْكُمْ مَنْ يُرَدُّ إِلَى
أَرْذَلِ الْعُمُرِ لِكَيْلًا بَعْلَمَ مِنْ بَعْدِ عِلْمٍ شَيْئًا وَتَرَى الْأَرْضَ هَامِدَةً فَإِذَا
أَنْزَلْنَا عَلَيْهَا الْمَاءَ اهْتَرَّتْ وَرَبَتْ وَأَنْبَتَتْ مِنْ كُلُّ زَوْجٍ بَهِيجٍ

### صدق الله العظيم

سورة الحج الآية 5

## **Dedication**

To my lovely parents

My sisters and brothers

Who have always been the candles of

my life

And the wind beneath my wings until I complete this work

#### Acknowledgment

Primary my prays and thanks should be to Allah, the almighty most gracious and most merciful, who grated me the serenity, means of strength and patience to accomplish this work.

I deeply indebted to my supervisor **Dr. Malik Hassan Ibrahim Elfadni** for his valuable help and guidance during this study, I'm also grateful to his keen interest, patience assistance and invaluable advice.

My appreciation is extended to all the medical staff in Heglig hospital and staff of National Health Laboratory department of haematology.

My special thanks are expressed to my colleagues Mohammed A. A. Allah, Alla Eldeen and Razan M. A. Elmotalab and to all my friends who have been very supportive throughout this work.

#### **Abstract**

This study was hospital analytical, descriptive and cross-sectional conducted to determine the sickle cell disease and sickle cell trait frequency in Sudanese patient in Heglig area in Western of Southern Kordofan state in the period between November 2008 to February 2009.

One hundred (100) patients were informed about the study and agreement for participation was obtained. A venous blood of 2.5 ml was collected in EDTA containers and investigated for sickle cell disease and sickle cell trait, a complete blood count (CBC), sickling test and haemoglobin electrophoresis were carried out.

Fully automated haematological analyzer (Sysmex Kx 21N), Electrophoresis tank and power pack were used for analysis and statistical package for social sciences (SPSS) computer program version 13 was used for data processing.

The mean age of the sickle cell disease patients was (7.92). The results showed that percentage of sickle cell trait and sickle cell disease were (52) and (14) respectively. The sickling test showed that (71%) of the study population were negative sickling test, the remain (29%) were positive.

Haemoglobin level, total erythrocyte and packed cell volume of the sickle cell disease patients were (6.59), (2.43) and (22.02), respectively.

There were no significant differences in sickle cell disease and normal individuals in mean cell haemoglobin concentration (29.80), mean cell volume (92.50) and mean cell haemoglobin (26.48) (p < 0.095, p < 0.226 and p < 0.400 respectively,

The total leukocytes (20.4) p < 0.000) and platelets count (280.78) p < 0.044) were significantly elevated in sickle cell disease when compared with normal individuals.

In conclusion, sickle cell anaemia is highly frequent in the studied area, which is most likely due to the consanguineous marriages.

#### 

هذه دراسة م قطعية, وصفية وتحليلية. تم إجراؤها لتحديد تردد مرض فقر الدم المنجلي وحاملي مرض فقر الدم المنجلي في الفترة ما مرض فقر الدم المنجلي في المرضي السودانيين في منطقة هجليج غرب ولاية جنوب كردفان في الفترة ما بين نوفمبر 2008 إلى مارس 2009.

تم إعلام مائة (100) مريض بأهداف البحث وأخذت مواف قتهم, ثم أخذت مائة عينة دم , 2.5 مل ethylene-diamine-tetra-acetic acid) من كل مريض في حاويات تحتوي علي مانع التجلط

EDTA))). تم إجراء اختبارات تعداد الدم الكامل, اختبار التمنجل و الرحلان الكهربائي للهمغلوبين.

تم استخدام جهاز (Sysmex) رقم (Kx 21N) يعمل أوتوماتيكيا, جهاز الرحلان الكهربائي للهمغلوبين و برنامج الحزم الإحصائية للعلوم الاجتماعية نسخة رقم 13 للتحليل الإحصائي.

و قد أظهرت النتائج الإحصائية أن متوسط أعمار مرضي ف قر الدم المنجلي (7.92). ونسب تردد مرضي ف قر الدم المنجلي وحاملي المرض 14%, 52% علي التوالي. أما اختبار التمنجل أظهر أن (71%) أظهروا نتيجة سالبة, أما (29%) أظهروا نتيجة موجبة.

وُجد أن مستوي الهمغلوبين, مجموع تعداد خلايا الدم البيضاء وحجم الخلية المحشوة لمرضي فقر الدم المنجلي (6.59), (22.02) و (22.02) على التوالي.

كما لم تُوجد فروق ذات دلالة إحصائية في متوسط تركيز الهمغلوبين للخلية (29.80) p < 0.095), متوسط حجم الخلية (p < 0.226 (92.50) ومتوسط همغلوبين الخلية (p < 0.400 (26.48)).

(p < 0.000~(20.41) ورُجدت زيادة ذات دلالة إحصائية في مجموع تعداد خلايا الدم البيضاء (p < 0.000~(20.78)) م قارنةً بأفراد أصحاء.

وُجد أن تردد فقر الدم المنجلي وحاملي مرض فقر الدم المنجلي عالي في المنطقة التي تمت دراستها, وهذه الخلاصة مردها لزواج الأقارب المنتشر في تك المنطقة.

### **List of Tables**

NO	Name of the tables	Page
1-1	Normal red blood cell values.	3
1-2	The classification of anaemia that based on red cell indices.	4
1-3	The classification of anaemias based on morphology (erythrocyte	4
	size).	
1-4	The common haemoglobinopathies.	15
1-5	The prevalence of haemoglobinopathies and thalassemia among	17
	Blacks.	
1-6	The Areas of high prevalence of sickle mutation.	21
1-7	The sickling syndromes.	22
1-8	The common abnormal haemoglobin.	22
1-9	The symptomatology and Clinical Manifestations of Vaso-	34
	occlusive Crisis.	
1-10	The presenting symptoms of acute chest syndrome.	37
1-11	The differential diagnosis in sickle cell syndromes.	41
3-1	The mean and Std. deviation of age among the study population	52
	group.	
3-2	The age groups among the study population group.	52
3-3	The full blood count for the study population group.	52
3-4	The sickling test among the study population group.	53
3-5	The full blood count for the sickle cell trait (Hb AS) group.	53
3-6	The full blood count for the sickle cell disease (Hb SS) group.	54
3-7	The full blood count in normal group (Hb AA), sickle cell trait	54
	(Hb AS) and sickle cell disease (Hb SS) groups.	
3-8	The full blood count and age in normal group (Hb AA) and sickle	55
	cell trait (Hb AS) group.	
3-9	The full blood count and age in normal group (Hb AA) and sickle	55
	cell disease (SS) group.	

## List of Figures

NO	Name of the Figures	Page
1-1	The location of Sudanese tribes and tribes groups where some	3
	genetics studies were performed.	
1-2	The CBC and reticulocyte index is used to classify anaemia	8
	morphologically.	
1-3	The Functional classification of anaemia.	8
1-4	The genetic control of human haemoglobin production in	12
	embryonic, fetal and adult life.	
1-5	The globin chain production and development.	13
1-6	The haemoglobin structure.	14
1-7	The geographic distribution of haemoglobinopathies.	16
1-8	The Distribution of malaria in southern Europe, southwest Asia,	21
	and Africa.	
1-9	How sickle cell genes are inherited.	23
1-10	The endothelial red cell adhesion and vaso-occlusion in sickle	30
	cell disease.	
1-11	The pathophysiology of sickle cell disease	33
1-12	The clinical problems of sickle cell disease by age.	36
3-1	The gender among the study population.	51
3-2	The haemoglobin AS, SS and AA among the study population	53
	group.	
3-3	The total erythrocytes (10 <sup>6</sup> / µl) in normal group (Hb AA), sickle	56
	cell trait (Hb AS) and sickle cell disease (Hb SS) groups.	
3-4	The haemoglobin (g/dl) in normal group (Hb AA), sickle cell trait	56
	(Hb AS) and sickle cell disease (Hb SS) groups.	
3-5	The mean cell hemoglobin concentration (g/dl)in normal group	57
	(Hb AA), sickle cell trait (Hb AS) and sickle cell disease (Hb SS)	
	groups.	
3-6	The mean cell volume (fl) in normal group (Hb AA), sickle cell	57
	trait (Hb AS) and sickle cell disease (Hb SS) groups.	
3-7	The packed cell volume (%) in normal group (Hb AA), sickle	58
	cell trait (Hb AS) and sickle cell disease (Hb SS) groups.	
3-8	The mean cell haemoglobin (pg) in normal group (Hb AA),	58
	sickle cell trait (Hb AS) and sickle cell disease (Hb SS) groups.	
3-9	The total leukocytes (10 <sup>3</sup> /µl) in normal group (Hb AA), sickle cell	59
	trait (Hb AS) and sickle cell disease (Hb SS) groups.	
3-10	The platelet count (10³/µl) in normal group (Hb AA), sickle cell	59
	trait (Hb AS) and sickle cell disease (Hb SS) groups.	

### List of abbreviations

AVN	Avascular necrosis
CBC	Complete blood count
CD 64	Cluster differentiation 64
CD 36	Cluster differentiation 36
gp Ib	Glycoproteins Ib
gp IX	Glycoproteins IX
gp V	Glycoproteins V
G-6-PD	Gglucose-6-phosphate dehydrogenase
FN	Fibronectin
Hb	Haemoglobin
Hb S	Haemoglobin S
Hb A	Haemoglobin A
Hb E	Haemoglobin E
Hb F	Haemoglobin F
HPFH	Hereditary persistence of fetal haemoglobin
LDH	Lactate dehydrogenase
MCV	Mean cell volume
MCH	Mean cell haemoglobin
MCHC	Mean cell haemoglobin concentration
$O_2$	Oxygen
PCV	Packed cell volume
RBC	Red blood cell.
SCD	Sickle cell disease
SPSS	Statistical package for social sciences
TWBCs	Total white blood cells count.
С	
TRBCsC	Total red blood cells count.
TSP	Thrombospondin
VCAM-1	Vascular cell adhesion molecule 1
vWF	von Willebrand factor
WBC	White blood cell.
2,3-DPG	2,3 diphosphoglycerate

### **List of contents**

NO	CONTENTS	Page
آڍة		I
Dedication.		
Acknowledgment.		III
Abstrac	ct in English.	IV
Abstract in Arabic.		V
List of tables.		VI
List of	figures.	VII
List of	abbreviations.	VIII
List of	contents.	IX
	CHAPTER ONE	
	Introduction and literature review	
1-1	Introduction.	1
1-2		3
	Classification of anaemia.	5
	Clinical features of anaemias.	9
	Haemoglobinopathy.	11
1-3-1		11
	Structure and function of haemoglobin.	11
1-3-3	Geographical distribution and prevalence of	15
	haemoglobinopathies.	
1-3-4		18
	General introduction sickle cell anaemia.	19
1-4-1	0 0 1	20
	anaemia.	
	Type of sickle cell anaemias.	22
	The inheritance of sickle cell anaemias.	23
1-4-4	Pathophysiology of sickle cell disease.	24
	Clinical features of sickle cell disease.	31
	Laboratory features.	39
1-5	Hemoglobin S syndromes.	39
1-5-1	Sickle cell trait.	39
1-5-2	Haemoglobin SC disease.	39
1-5-3	Sickle cell–b-thalassaemia.	39
1-5-4	Sickle cell anaemia with coexistent α-thalassaemia.	39
1-5-5	Haemoglobin S/hereditary persistence of fetal hemoglobin	39
	(Sickle cell–HPFH).	

1-5-6	Hemoglobin SE disease.	39		
1-5-7	Haemoglobin SD disease.	40		
1-5-8	Haemoglobin SO-Arab disease.	40		
1-5-9	Haemoglobin S Hb Lepore disease.	40		
1-6	Rationale.	42		
	Objective.	43		
	CHAPTER TWO			
	Methodology			
2-1	Study design.	44		
2-2	Study area.	44		
	Study population.	44		
2-3-1	1 0	44		
	inclusion criteria.	44		
2-3-3	exclusion criteria.	44		
	Sample size.	44		
		45		
2-5	Data analysis.	45		
2-6	Ethical consideration.	45		
2-6	Methodology.	45		
2-6-1	Method of sample collection.	45		
2-6-2	Method of automated analyzer system (Complete	46		
	haemogram).			
2-6-3	Method of Preparation and staining of blood films:	47		
2-6-4	Sickling Test.	48		
2-6-5	Electrophoresis method.	49		
CHAPTER THREE				
	The results			
3-	Result	51		
	CHAPTER FOUR			
	Discussion, Conclusion and Recommendation			
4-1	Discussion:	60		
4-2	Conclusion:	64		
4-3	Recommendation	64		
CHAPTER FIVE				
References				
References:		66		
Appendixes		71		