

Knowledge and experiences of parents with children affected by Sickle Cell Disease in Cape Town

by

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SUBMITTED TO THE UNIVERSITY OF CAPE TOWN

in partial fulfilment of the requirements for the degree

MSC (MED) GENETIC COUNSELING

Faculty of Health Sciences

UNIVERSITY OF CAPE TOWN

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Date of submission: 15 August 2015

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Abstract

Sickle Cell Disease (SCD) is an autosomal recessively inherited blood disorder that leads to a debilitating systemic illness. Although the disease was initially found predominantly in tropical and subtropical regions, SCD has now become a global health problem, due to migration of people from various countries with a high burden thereof. Consequently, the incidence of SCD in South Africa has increased dramatically over the last decade.

This study, which constitutes a minor dissertation in fulfilment of an MSc (Med) Genetic Counselling degree, aimed to explore the knowledge and understanding of SCD among parents of affected children in Cape Town as well as identify burdens associated with caring for a child with SCD. Furthermore, the study assessed opportunities to improve genetic counselling services available to parents and explored their attitude to preventive policies.

A phenomenological approach was used to conduct this research. Seventeen semi-structured interviews were conducted with the biological parent of a child attending the Red Cross War Memorial Children's Hospital Haematology Clinic. Participants were selected using both purposive and convenience sampling methods. Data collected during these interviews were analysed using thematic content analysis.

Themes and relevant sub-themes were identified and grouped into three categories: knowledge and understanding; experiences and burdens; and attitude toward preventative policies. While the majority of participants had some knowledge of SCD, several misconceptions were discovered, often relating to participants' prior knowledge of the disease. A number of burdens experienced by participants were revealed, with both practical and psychosocial implications. Finally, it was found that the majority of participants supported all methods of screening for SCD, regardless of whether they would make use of the screening services themselves.

Findings of this study provide valuable insights on the subject of experiences of parents of children affected with SCD as well as the potential role of genetic counselling services. This study contributes towards improving understanding and subsequent services provided to individuals raising a child affected with Sickle Cell Disease.

Acknowledgements

I would like to thank Professor Ambrose Wonkam for giving me the chance to do this project, introducing me to the fascinating world of Sickle Cell Disease, and his invaluable feedback. Thank you to Dr Jantina de Vries for enriching and informing my experience with qualitative research and to Ms Nakita Laing for being across the hall and willing to help and support at any time. Thanks to you I was able to (mostly) enjoy the research process.

I would also like to thank Dr Tina-Marié Wessels and Dr Jacquie Greenberg for the wonderful opportunity to do this course. A special thank you to my fellow students, Gill and Tarryn, for their willingness to listen, encourage, support, and buy coffee.

And lastly, thank you to my husband, without whom this thesis, like so many things I do, would never ever have been possible.

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List of Terms and Abbreviations

AIDS	Acquired Immune Deficiency Syndrome
Allele	Each of two or more alternative forms of a gene that arise by mutation and are found at the same place on a chromosome
DNA	Deoxyribonucleic acid
Genotype	The combination of alleles located on homologous chromosomes that determines a specific characteristic
Haemoglobinopathies	Inherent disorders that result in abnormal structure of one of the globin chains of the haemoglobin molecule
HbA	Adult haemoglobin
HbF	Foetal haemoglobin
HbS	Sickle haemoglobin variant
Heterotetramer	A protein containing four non-covalently bound subunits
Heterozygous	Having different alleles at a given locus on a pair of chromosomes
HIV	Human Immunodeficiency Virus
Homozygous	Having identical alleles at a given locus on a pair of chromosomes
Incidence	Number of new cases of a disease that develop in a given period of time
Ischemia	An inadequate blood supply to an organ due to blockage of blood vessel leading to that organ

<i>Plasmodium falciparum</i>	Parasite that causes malaria
Prevalence	The number of cases of a disease that are present in a particular population at a given time
RCWMCH	Red Cross War Memorial Children's Hospital
SA	South Africa
SCA	Sickle Cell Anaemia
SCD	Sickle Cell Disease
SNP	Single Nucleotide Polymorphism
UK	United Kingdom
USA	United States of America
WHO	World Health Organization

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Chapter 1: Introduction and Literature Review

1.1 Introduction

This research was conducted as part of the requirements for completion of the MSc (Med) Genetic Counselling degree, and explored the knowledge, understanding and experiences of parents of children affected with SCD in Cape Town. Considering the rise in patients with SCD in the South African healthcare context, discussed in more detail in section 1.2.6, the public health service will need to adapt and adjust in order to provide the necessary services. However, there is currently no data on what people with SCD in Cape Town experience, both with regards to caring for a child with SCD and in connection to the health services they receive. The nature of the burdens experienced by parents in South Africa have also not been previously investigated.

This first chapter comprises a review of the literature on the epidemiology of Sickle Cell Disease (SCD), the genetics of the condition, and the clinical description thereof. The chapter will also consider the psychosocial burdens experienced by caretakers of children affected by SCD as well as various SCD screening options and how the uptake thereof could influence disease incidence. Finally, the review will take a brief look at SCD in South Africa (SA) generally and Cape Town specifically, and discuss disease frequency and available services. Altogether, this chapter will serve as a background introduction for this study, which aims to explore knowledge and experiences of parents with children affected with SCD in Cape Town. These aims are introduced in more detail at the end of the chapter.

Chapter Two describes the methodological approach used in this study, while Chapters Three to Five report and discuss the results. Each of these chapters present a combination of the results and discussion on the findings, as grouped by subject. Chapter Six provides a summary of the main findings, as well as recommendations based on those findings.

1.2 Literature review

1.2.1 Epidemiology of Sickle Cell Disease

Sickle Cell Disease (SCD) was initially described by a Dr Herrick, who first noted the characteristic sickle shaped red blood cells in 1910 (Rees, Williams, & Gladwin, 2010), yet it has been known in certain parts of Africa since well before the 20th century (Makani, Ofori-

Acquah, Nnodu, Wonkam, & Ohene-Frempong, 2013). SCD is an autosomal recessive haemoglobinopathy, characterised by the presence of sickle haemoglobin (HbS), a haemoglobin variant caused by the amino acid substitution of valine for glutamic acid at the sixth position of the β -globin chain. This substitution is the result of a single nucleotide replacement, adenine in place of thymine, within the β -globin gene (Frenette & Atweh, 2007). With the β -globin gene in the deoxygenated state, HbS has the tendency to polymerise, distorting the red blood cells into the distinctive sickle shape (Rees *et al.*, 2010). These deformed and rigid blood cells can interfere with normal blood flow in microcirculation and induce ischemia in tissues distal to the vascular blockage. This vasoocclusion is one basis for the observed SCD complications, which include painful crises, susceptibility to infection, stroke, and chronic organ damage (Bartolucci & Galactéros, 2012), which will be discussed in more detail shortly.

Inherited haemoglobin disorders, such as SCD, were originally most commonly found in tropical regions (Piel *et al.*, 2013), such as sub-Saharan Africa and central India. This origin in tropical areas is most likely due to the resistance to malaria conferred by the heterozygous state (Chakravorty & Williams, 2014). Carriers of sickle cell trait, or those with only one copy of the gene mutation, have lower rates of mortality from malaria infection when compared to non-carriers, giving them an evolutionary advantage. The sickle haemoglobin variant (HbS) seems to lessen the risk of infection by *Plasmodium falciparum*, the parasite responsible for causing malaria (Piel *et al.*, 2010). Although malaria is often fatal to someone affected with SCD, the protection appears to work in a dose-dependent manner, which means people with Sickle Cell Anaemia (SCA), who are homozygous carriers of HbS, have an even lower risk of being infected with malaria than a heterozygous carrier (Ashley-Koch, Yang, & Olney, 2000). The mechanism of this protective effect is not fully understood, but is thought to include both innate and immune-mediated mechanisms (Rees *et al.*, 2010).

Whatever the cause may be, it is this protective effect that is believed to be responsible for the high frequency of the HbS variant (Piel *et al.*, 2010). If carriers of the HbS variant are less likely to become infected with malaria in areas where that disease is prevalent, they are consequently more likely to reach reproductive age (Makani *et al.*, 2013). In fact, the HbS mutation has been described as the archetypal example of natural selection in humans (Chakravorty & Williams, 2014). As a result of this, populations where malaria is or was endemic can reach HbS carrier frequencies of up to 40% (Modell & Darlison, 2008; Weatherall & Clegg, 2001).

However, even if malaria as an elective force were to be removed it would take many generations for the gene frequencies to fall significantly (Weatherall, 2011). Moreover, despite the fact that SCD originated in malaria infested tropical regions, the HbS allele has now spread globally (Figure 1.1) due to human migration (Piel *et al.*, 2013). As a result inherited haematological disorders, or haemoglobinopathies, are now the most common monogenic disease worldwide (Modell & Darlison, 2008; Piel *et al.*, 2013).

Despite this fact, determining the exact global prevalence of SCD can be difficult, as it requires the accurate diagnosis and registration of all SCD affected children at birth.

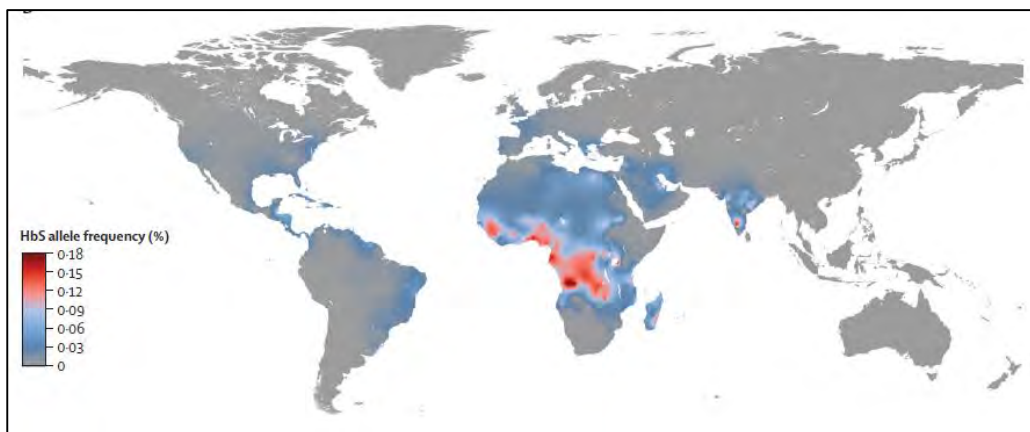


Figure 1.1: Predicted global distribution of HbS allele frequency (Piel *et al.*, 2013).

While this is efficiently done in countries such as the United States of America (USA) and the United Kingdom (UK), it is limited in African countries, which make up 75% of the global SCD population (Makani *et al.*, 2013). To overcome this problem, an alternative method of determining disease prevalence is to use the prevalence of the heterozygous carrier state to calculate the expected birth rates based on the gene frequency and Hardy-Weinberg equation (Makani *et al.*, 2013; Piel *et al.*, 2013). It has been found that 5.2% of the world population carry a haemoglobin variant, of which 40% carry the HbS variant that contributes towards SCD (Modell & Darlison, 2008). Calculations made with these frequencies have estimated that worldwide, more than 300 000 children are born with SCD every year (Chakravorty & Williams, 2014; Makani *et al.*, 2013; Modell & Darlison, 2008).

As a result of this high disease incidence, haemoglobinopathies present a health burden comparable to that of communicable diseases (Weatherall, 2008). This is particularly true in the

developing world where diagnosis and treatment is not always readily available. The burden of SCD has been further highlighted by the high birth prevalence, resulting in the World Health Organization (WHO) recommending SCD as a public health priority in 2006 (Makani *et al.*, 2013).

Moreover, as SCD can be cost-effectively controlled through programs that integrate treatment, carrier detection and genetic counselling, as discussed later, the WHO has also recommended global development of these services to combat the high disease burden (Modell & Darlison, 2008).

1.2.2 Genetics of Sickle Cell Disease

Understanding the underlying genetics of SCD is vital, as genotype is the most important risk factor and predictor of SCD disease severity (Ashley-Koch *et al.*, 2000), which can vary considerably. Genotype will be an indication of the types of haemoglobin present in the blood of an affected person, which in turn will be an indication of severity.

Normal Haemoglobin

Normal adult haemoglobin (HbA) are heterotetramers, consisting of α - and β -like globin chains, each carrying an iron containing molecule called heme (Figure 1.2). This molecule enables the protein to carry out its major function of delivering oxygen from the lungs to peripheral tissue throughout the body (Thom *et al.*, 2013).

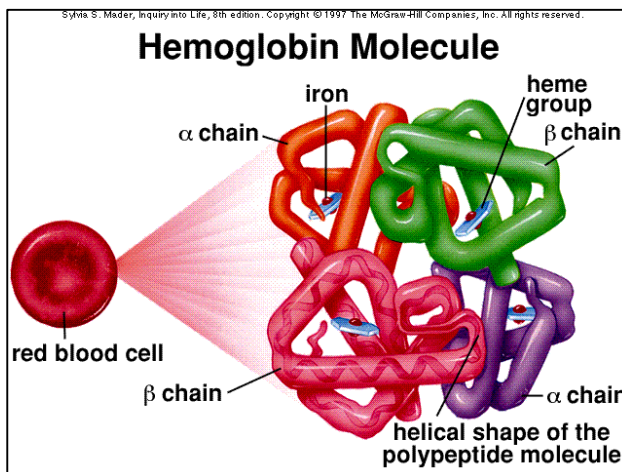


Figure 1.2: Structure of HbA (Mader, 1997)

The instructions for making haemoglobin come from the globin family, a group of genes involved in oxygen transport (Ashley-Koch *et al.*, 2000). The genes found in this group are developmentally regulated (Figure 1.3), so that specific types of globin are present at specific times throughout human development (Thom *et al.*, 2013). The type of globin (α , β , δ , γ , ϵ , or ζ) present will in turn change the structure of human haemoglobin found during embryonic, foetal and adult life (Weatherall & Clegg, 2001). For example, adult (HbA) and foetal haemoglobin (HbF) consist of α -globin chains combined with β -, γ -, or δ -globin chains, while embryonic haemoglobin is made up of α -like chains combined with γ - or ϵ -globin chains (Makani *et al.*, 2013; Weatherall & Clegg, 2001).

During foetal life, HbF is the predominant type of haemoglobin present in the blood, but this changes around birth when a switch occurs from HbF to HbA gene expression, at which time HbF is gradually replaced by HbA, and by 6 months of age it is the primary haemoglobin found in the blood (Sankaran & Orkin, 2000; Thom *et al.*, 2013). In a person affected with SCD, this replacement of one type of haemoglobin with another will mark the start of symptoms, as HbF will in their case not be replaced by HbA, but rather by HbS.

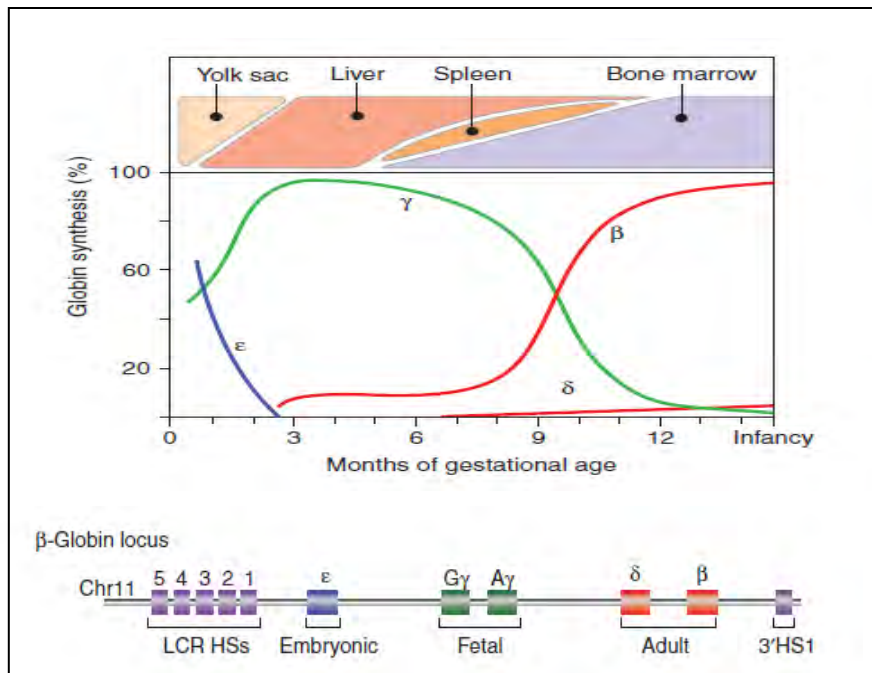


Figure 1.3: HbF to HbA switch. This figure illustrates the timing of the switch from HbA to HbF. The top section shows the site and level of various types of globin as expressed at specific times during development. The colours used correspond to those used in the bottom section in a depiction of the developmental groups of genes found on the β -globin locus (Sankaran & Orkin, 2000).

Thalasseмии and Sickle cell Disease

Haemoglobin gene variants are present in all populations of the world at low frequencies, due to spontaneous mutations (Modell & Darlison, 2008). These mutations fall into two main groups: thalasseмии and structural variants. Thalasseмии lower the production of globin chains resulting in red blood cells of reduced quantity, while structural variants lead to changes in amino acid sequence and produces abnormal haemoglobin, resulting in red blood cells of reduced quality (Modell & Darlison, 2008). This is the case in SCD, where the quality of the red blood cell is compromised by a single nucleotide mutation in the β -globin gene, called the β s-mutation (Ashley-Koch *et al.*, 2000). Because of this mutation and the resulting substitution of valine for glutamic acid, a hydrophobic motif in the deoxygenated HbS tetramer is caused, leading to a bond being formed between two β -globin chains (β 1 and β 2) of two haemoglobin molecules (Rees *et al.*, 2010). This crystallization creates a polymer nucleus, which in turn causes cellular stress by disrupting flexibility and promoting cellular dehydration (Rees *et al.*, 2010).

The most common form of SCD is sickle cell anaemia (SCA), which occurs as a result of the homozygous inheritance of the β s-mutation. In populations of African origin, SCA accounts for 70% of cases of SCD (Rees *et al.*, 2010). SCA, like SCD, is inherited in an autosomal recessive pattern. This means that in order for the disease to be expressed, two copies of the disease causing mutation had to have been inherited by the affected individual, one from each parent (Ashley-Koch *et al.*, 2000). The primary haemoglobin present in the red blood cells of individuals suffering from SCA is HbS (Edwards *et al.*, 2005).

SCD can, however, also occur as a result of the inheritance of HbS in combination with any one of an extensive range of other haemoglobin mutations in individuals who are then referred to as compound heterozygotes (Chakravorty & Williams, 2014). In addition to one copy of the HbS gene variant, compound heterozygotes also carry a copy of another β -globin gene variant, such as haemoglobin C (HbC), which results in Sickle Haemoglobin C Disease, or Hb β -thalassemia, which results in Sickle Cell/ β -thalassemia (Frenette & Atweh, 2007). Compound heterozygotes produce a mixture of variant haemoglobins, resulting in variable disease severity (Steinberg, 2009).

If a person inherits only one copy of the β s-mutation in addition to one normal gene copy, that person is said to be a carrier. Carriers produce a mixture of HbS and normal HbA. In the case of

SCD, the carrier state is also often referred to as sickle cell trait. Individuals with sickle cell trait are not affected by SCD but it is important for these individuals to understand the reproductive implications (Treadwell, McClough, & Vichinsky, 2006). Children born to two parents with sickle cell trait have a 1 in 4 (25%) chance of being affected with SCD and a 1 in 2 (50%) chance of being a carrier themselves.

In addition to genotype, a further genetic influence on SCD disease severity is the β -globin haplotype. A haplotype refers to a combination of DNA markers observed on a particular chromosome. On chromosome 11, near the β -globin gene, there is a cluster of several other globin genes. This region is referred to as the β -globin cluster region and as the DNA markers found in this region are highly variable, many haplotypes exist. However, only five of these haplotypes are associated with the HbS mutation, namely: Benin; Senegal; Central African Region/Bantu; Cameroon and Arab-Indian (Bitoungui, Pule, Hanchard, Ngogang, & Wonkam, 2015). The haplotypes are named for the geographical regions where they were first observed, and because they are population specific (Figure 1.4), it is theorised that the β s-mutation arose independently in each of these populations (Rees *et al.*, 2010). The β -globin haplotypes are associated with differences in clinical severity, which will be discussed in more detail in the section on pathophysiology and clinical symptoms of SCD.

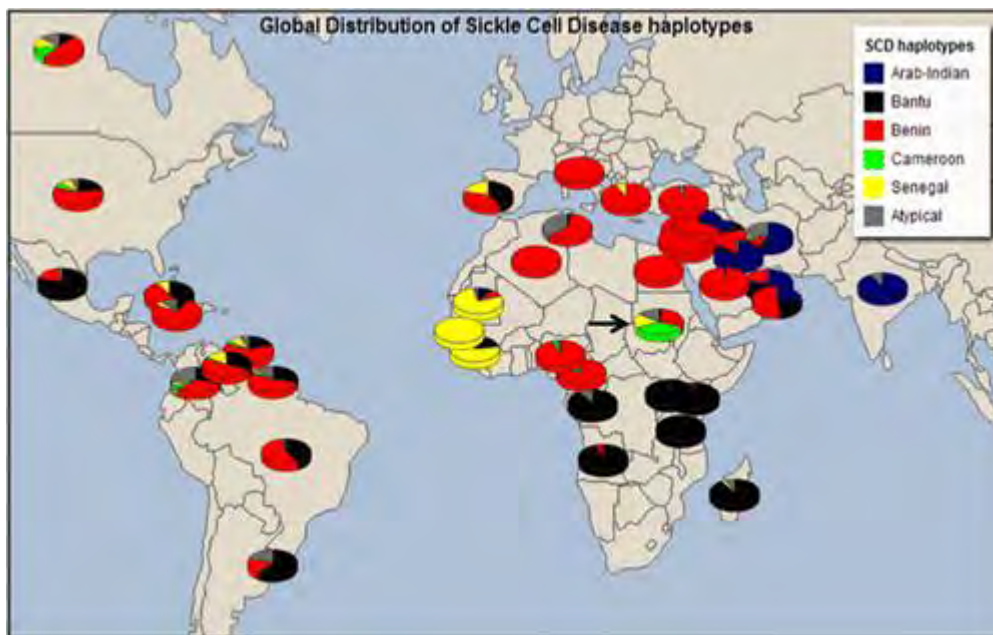


Figure 1.4: Global distribution of haplotypes among various world SCD populations (Bitoungui *et al.*, 2015).

1.2.3 Pathophysiology and clinical manifestation of Sickle Cell Disease

Sickle cell disease is essentially a multisystem disorder, affecting practically every organ system of the body (Makani *et al.*, 2013). It is caused by an abnormality of the red blood cells that arises due to the abnormal amino acid found in the β -globin chain. Unlike HbA, HbS forms long, insoluble polymers when deoxygenated (Bartolucci & Galactéros, 2012), which causes the red blood cells to lose their characteristic biconcave disc shape. Instead, the red blood cells take on the irregular sickle or crescent shape for which the disorder is named.

The clinical course of SCD often results in noticeable physical morbidity, as it includes periods of acute and chronic haematological crisis, which can at times lead to early death (Edwards *et al.*, 2005). Illness severity can vary, and is associated with a range of acute and chronic health complications including pain crises, acute chest syndrome, cerebrovascular accidents and splenic and renal dysfunction (Rees *et al.*, 2010). Makani *et al.* (2013), divided the considerable clinical consequences of SCD into four categories: haemolysis and haematological complications; vasoocclusion; end organ dysfunction; and infection.

Haemolysis and Haematological Complications

Red blood cells with a sickled shape are susceptible to haemolysis, or rupturing, and show an average lifespan that is 6 -9 times shorter than that of normal red blood cells. This causes individuals with SCD to develop chronic haemolytic anaemia that will be present throughout their life (Rees *et al.*, 2010). In addition to the chronic anaemia, individuals affected with SCD may also intermittently experience acute episodes of reduction in haemoglobin, known as an anaemic crisis (Makani *et al.*, 2013). An anaemic crisis is characterized by a sudden fall in steady haemoglobin as well as increased reticulocytes (immature red blood cells) and exaggerated hyperbilirubinemia (jaundice). The latter causes a yellow discoloration of the skin and sclera, often seen in persons affected with SCD. While chronic anaemia itself is usually relatively moderate, it can lead to various secondary complications, for example gall bladder disease due to high levels of bilirubin. Furthermore, the presence of any viral infection can cause a substantial reduction in red blood cell production, which, in turn, leads to a more severe, life threatening anaemia, called an aplastic crisis (Ashley-Koch *et al.*, 2000).

Vasooocclusion

Besides the abnormal shape of the red blood cells causing anaemia, sickled cells also have a tendency to adhere to the walls of blood vessels, thereby clogging the vessels and preventing normal blood flow (Chakravorty & Williams, 2014). This blocking of the blood vessels is called vasooocclusion (Figure 1.5) and the resulting decrease in blood flow disrupts the flow of oxygen to organs and tissues throughout the body. This can often result in considerable tissue damage, for example acute splenic sequestration (Makani *et al.*, 2013), which occurs as a result of sickled red blood cells being trapped inside the spleen. Acute splenic sequestration is clinically characterized by a sudden increase in splenic size, which causes reduced splenic function, and can necessitate the removal of the spleen. In certain life threatening instances it can also be associated with severe anaemia and hypovolemic shock (Chakravorty & Williams, 2014).

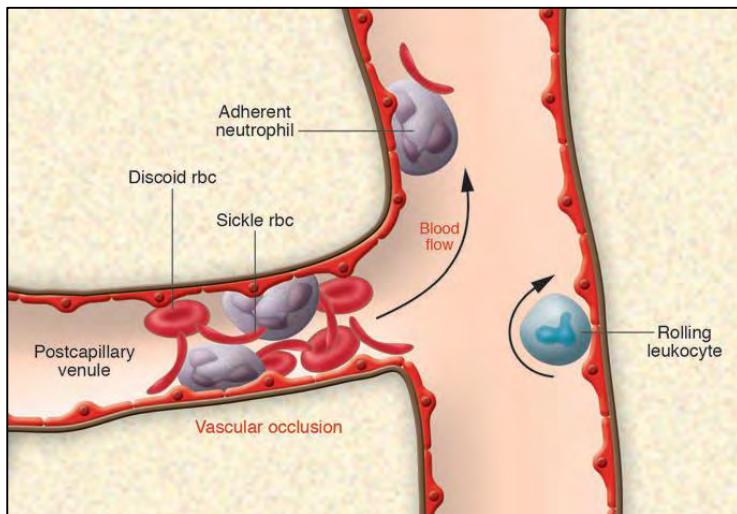


Figure 1.5: Sickle Cell Vasooocclusion (Frenette & Atweh, 2007).

Vasooocclusion is also thought to be the underlying cause of severe painful episodes suffered by patients with SCD (Edwards *et al.*, 2005). These pain crises are considered the hallmark of SCD and contribute considerably to its morbidity. They are episodes of intense muscoskeletal pain, usually affecting the arms, legs, back, abdomen, chest and head of affected individuals (Ashley-Koch *et al.*, 2000; Makani *et al.*, 2013). While it has been suggested that pain crises may be brought on by an increase in the ratio of sickle to regular shape red blood cells, an increase in barometric pressure or nutritional and stress factors (Edwards *et al.* 2005), the exact trigger thereof is usually unpredictable (Chakravorty & Williams, 2014). Pain crises are the most

common cause of hospitalisations among SCD affected individuals, irrespective of the numerous other physiological complications associated with the disease (Rees *et al.*, 2010). Despite this fact, more than 80% of pain crises occur and are managed in a home setting through caregivers providing medication or comfort measures (Ely, Dampier, Gilday, O'Neal, & Brodecki, 2002). This illustrates the extensive presence of pain throughout the lives of children and families affected with SCD.

As well as being very painful in itself, these pain crises also contributes greatly to depression, anxiety and other psychiatric disturbances found in persons suffering from SCD (Edwards *et al.*, 2005). These psychiatric disturbances are frequently associated with a diminished ability to cope with intense pain, and in turn help continue the cycle of intensifying pain with significant life dysfunction and functional disability that is often marked in SCD. Because of the substantial chronic pain component and its impact on psychosocial well-being, it has been suggested that SCD is better conceptualised as a disease with psychosocial as well as physiological complications (Edwards *et al.*, 2005).

End Organ Dysfunction

End organ dysfunction refers to damage occurring in major organs fed by the circulatory system. Several major organs of individuals with SCD are eventually damaged, with this occurrence becoming particularly evident due to increased life expectancy following suitable treatment (Bartolucci & Galactéros, 2012)

Several medical complications can result from tissue damage throughout the body and includes delayed growth and sexual maturity; acute and chronic pulmonary dysfunction; sickle cell retinopathy; and dermal ulcers (Edwards *et al.*, 2005). However, the most particularly affected organs are the brain and lungs. Cerebrovascular complications in SCD are a well described event and occur as a result of the brain being affected by obstructed blood flow. The complications can include temporary ischemic attacks, ischemic strokes, and haemorrhagic strokes that are sometimes associated with seizures (Chakravorty & Williams, 2014). Lungs are affected through an illness called acute chest syndrome, which is a life threatening pneumonia-like illness characterized by the development of new pulmonary infiltrates involving at least one lung segment (Rees *et al.*, 2010). Both these examples occur frequently in persons affected with SCD and are important risk factors for death (Platt *et al.*, 1994).

Infection

Individuals with SCD are incredibly susceptible to bacterial infection and, in the absence of prophylaxis, infections are thought to be the leading cause of clinical events associated with increased mortality. This increased susceptibility could arise due to factors related to immunity, both cellular and humeral, as well as intrinsic complications of SCD or the treatment thereof (Ashley-Koch *et al.*, 2000; Makani *et al.*, 2013). For instance, end organ damage can leave certain sites vulnerable to infection, while chronic haemolysis leads to high bone marrow turnover, which results in increased vulnerability to viral infections (Makani *et al.*, 2013). Furthermore, reduced splenic function (discussed above) is well known for causing immune deficiencies, as the spleen plays an important part in the human immune system. It has been suggested that by 5 years of age, 94% of individuals with SCA are functionally asplenic (Makani *et al.*, 2013), thereby losing its protection against infection. All these factors contribute towards a significant increase in immune vulnerability among affected individuals.

Mortality in Sickle Cell Disease

SCD is associated with significant mortality owing to a wide variety of causes. Among children and adolescents, death often occurs as a result of infection or cerebrovascular incidents (Ashley-Koch *et al.*, 2000). Among adults the cause of mortality is more varied, and can include acute splenic sequestration and acute chest syndrome (Grosse *et al.*, 2011; Makani, Cox *et al.*, 2011)

Irrespective of the cause, the result is a significant decrease in life expectancy among individuals with SCD. In the USA the average life expectancy of affected individuals is 42 and 48 years for men and women respectively. This is close to 30 years below that of the normal population (Platt *et al.*, 1994). However, life expectancy is greatly influenced by medical care as well as psychosocial processes (Edwards *et al.*, 2005), and can be considerably lower in developing countries. Studies done in Nigeria reported childhood mortality of up to 90% among children suffering from SCD, but recent estimates have suggested a decrease in mortality rates, which are more likely in the range of 50% by 20 years of age. This is similar to rates found in the USA in the early 1960s, which has since been significantly reduced with the help of early diagnosis and comprehensive treatment (Grosse *et al.*, 2011; Makani, Cox *et al.*, 2011).

Diagnosis of Sickle Cell Disease

Diagnosing SCD requires the analysis of the haemoglobin found in the red blood cells. In other words, determining the presence of HbS and the absence, or significant reduction of HbA (Chakravorty & Williams, 2014). The three tests most widely used for this purpose are haemoglobin electrophoresis, isoelectric focusing, and high performance liquid chromatography. All have a 93-100% sensitivity and 99-100% specificity, and can be performed on any blood samples, including umbilical cord blood or blood from a heel prick collected shortly after birth (Ashley-Koch *et al.*, 2000).

Alternatively, DNA testing can be performed in order to examine the disease causing mutation in the β -globin gene itself. This method becomes particularly necessary when no blood sample is available, as would be the case during prenatal testing. In such circumstances, DNA testing of the foetus can be done on a sample of the amniotic fluid or chorionic villus, in order to give an indication of whether the foetus will be affected or unaffected (Ashley-Koch *et al.*, 2000).

Disease Severity

There is considerable variability in terms of disease severity in SCD and, as mentioned in the review on SCD genetics, genotype is the most important indicator thereof. Individuals with SCA are most severely affected, followed by individuals with Sickle Cell/ β^0 thalassemia. Individuals affected with Sickle Cell Haemoglobin C Disease and Sickle Cell/ β^+ thalassemia tend to be less severely affected (Ashley-Koch *et al.*, 2000; Steinberg, 2009)

Persons diagnosed with SCA and Sickle Cell/ β^0 thalassemia have a higher rate of acute chest syndrome and pain crises than individuals with Sickle Haemoglobin C disease and Sickle β^+ thalassemia. Individuals with SCA also have the highest rate of cerebrovascular complications when compared to the other types of SCD. On the other hand, individuals with Sickle Haemoglobin C disease have an increased risk of thromboembolic complications, retinopathy and renal papillary necrosis in comparison to those with SCA. There is also a difference in life expectancy, with that of individuals affected with SCA being on average 20 years less than that of individuals with Sickle Haemoglobin C disease, most probably as a result of the difference in clinical severity between the two genotypes (Ashley-Koch *et al.*, 2000).

Besides genotype, other factors have also been identified as associated with SCD disease severity. These factors include level of HbF, β -globin gene cluster haplotype, and α -globin gene complement (Steinberg, 2009). HbF, if present at increased levels, can result in a less severe clinical form of SCD. It has been found that some individuals show a genetic predisposition to unusually high levels of HbF. This occurs as a result of over expression of γ -globin chains in certain adults. However, this hereditary persistence of HbF is very rare, and while the majority of affected individuals have varying levels of residual HbF it is not high enough to suppress severity (Ashley-Koch *et al.*, 2000; Steinberg, 2009).

The influence of the β -globin haplotype on disease severity is due to its impact on this variation in the level of HbA and HbF (Ashley-Koch *et al.*, 2000). Concentration levels of both HbA and HbF vary depending on haplotype, with genetic variants at three loci (BCL11A, HBS1L-MYB and the HBB cluster) accounting for 10–20% of HbF variation among SCD patients in the USA, Brazil (Lettre *et al.*, 2008) and the UK (Thein & Menzel, 2009). Initial studies in Tanzania and Cameroon have also shown that single-nucleotide polymorphisms (SNP) in the BCL11A loci are prevalent in both Tanzanian and Cameroonian patients, with significant association of these SNPs with HbF (Makani, Menzel *et al.*, 2011; Wonkam, Ngo Bitoungui, *et al.*, 2014). Of the three most common haplotypes, Senegal, Benin and Central African Republic, the first is associated with the least severe form of SCD. This is followed by the Benin haplotype, while the Central African Republic haplotype is associated with the most severe form of the disease (Steinberg, 2009).

Perhaps most importantly, levels of HbF is amendable to therapeutic manipulation, and certain therapies for SCD, such as hydroxyurea, have been developed using this principle and work by increasing the level of HbF in the blood of affected individuals (Sankaran & Orkin, 2000; Steinberg, 2009).

Finally, among individuals with SCD, coinheritance of α -thalassemia, an α -globin gene variant, seems to be protective against certain disease complications, specifically acute chest syndrome, anaemia and cerebrovascular accidents (Hsu *et al.*, 2003). On the other hand, this coinheritance increases susceptibility to other disease complications, such as pain crises (Embury *et al.*, 1982).

Despite this knowledge regarding the factors that can influence disease severity, predicting the progression and outcome of SCD from birth remains difficult (Rumaney *et al.*, 2014).

Treatment of Sickle Cell Disease

The main focus of the treatment of SCD involves clinical management of the symptoms, which can be divided into four approaches: supportive, symptomatic, preventative and abortive. The first is the most common, and involves assuring a balanced diet, adequate hydration and supplementation of folic acid. Symptomatic treatments are those targeted at alleviating specific SCD symptoms (Pule & Wonkam, 2014). For example, blood transfusions can relieve anaemia by increasing the level of normal haemoglobin present in the body and reducing the number of sickled blood cells in circulation (Makani *et al.*, 2013). The preventative approach is aimed at prohibiting certain disease complications, and includes pneumonia and influenza vaccinations and the use of hydroxyurea (Pule & Wonkam, 2014). Hydroxyurea is a cytotoxic drug that has been shown to be effective in reducing morbidity and improving survival among individuals with SCD, as well as reducing the frequency of pain crises and consequent hospital admissions (Makani *et al.*, 2013). While there might be additional benefits, including increased life expectancy and protection against cerebrovascular disease, the use of cytotoxic drugs are limited to patients with a severe clinical course due to fear of possible toxic effects (Rees *et al.*, 2010). The abortive approach is limited, with nitric oxide being the only accepted agent reported to completely stop chronic pain episodes in certain SCD patients (Pule & Wonkam, 2014).

A possible fifth approach also exists, not aimed at managing symptoms but at curing them, the ultimate goal for all genetic disorders (Pule & Wonkam, 2014). Haematopoietic stem cell transplantation remains the only potential cure for SCD but, due to high associated morbidity and mortality as well as the necessity of a suitably matched donor, it is only considered when serious complications have occurred (Carey, 2014; Oringanje, Nemecek, & Oniyangi, 2009). Gene therapy is promising and future developments might include the use of induced pluripotent stem cells as a source of haematopoietic progenitors for gene therapy (Rees *et al.*, 2010). This is not yet a reality though, and for the moment the difficulty of balancing short term treatment risks against long term disease complications remain a serious problem.

1.2.4 Psychosocial burdens experienced by Sickle Cell Disease caregivers

As mentioned above, SCD should be conceptualised as a disease with psychosocial as well as physiological components. The psychosocial components can be far reaching and therefore the effects of SCD are by no means limited to affected individuals. The role of caring for someone with SCD can cause personal and family conflicts, conflicts at work, trigger strain, stress,

embarrassment, fatigue and depression, thereby creating a burden for caregivers and impacting on their quality of life (da Silva *et al.*, 2012; Panepinto, O'Mahar, DeBaun, Loberiza, & Scott, 2005).

A number of studies have investigated the negative impact that SCD can have on the family and caregivers of individuals with SCD (Brown *et al.*, 2010; da Silva *et al.*, 2012; Moskowitz *et al.*, 2007; Wonkam, Zameyo Mba, *et al.*, 2014) and a variety of burdens have been identified, ranging from hospital visits that interfere with work commitments (van den Tweel *et al.*, 2008) to increased marital distress (Brown *et al.*, 2010).

This section will discuss some of the burdens experienced by caregivers of persons affected with SCD in more detail as well as their impact on the quality of life of the caregiver. For the sake of this discussion, burden can be defined as an objective or subjective impact, pressure or overload on caregivers, where subjective refers to attitudes and emotional reactions related to the experience of caring, and objective relates to changes in the daily life of caregivers (da Silva *et al.*, 2012). Quality of life, in turn, is connected to lifestyle and includes mental state, well-being and satisfaction of the caregiver or parent. These concepts are formed by knowledge and experience, through moral, religious and psychological principals as well as by social and cultural values (da Silva *et al.*, 2012). Together, these concepts shape a person's perspective of their own quality of life and whether it has increased or decreased in a given situation.

In a study conducted in the Netherlands, van den Tweel *et al* (2008) showed that caring for a child with SCD reduced the quality of life of caregivers, with an increase in depressive moods and reductions of happiness, vitality, cognitive function and sleeping. Moskowitz *et al* (2007) in a study in the USA found that 50% of SCD caregivers studied were at risk of clinical depression, compared to 19% of the control group, consisting of carers of healthy children. Such mood disturbances may be the result of a number of things, both physical and emotional. For example, sleep deprivation, as a result of night-time caregiving duties, can be a contributing factor to the decrease in vitality and happiness of parents caring for a child with SCD (van den Tweel *et al.*, 2008).

On an emotional level, the caregiver's sense of guilt and perception of how stigmatized the disease is can also contribute to feelings of depression (Moskowitz *et al.*, 2007). A Kenyan study on gendered experiences of stigma among families affected by SCD found stigmatisation to be frequently associated with the attribution of cause and subsequent blaming of the mother of

affected children (Marsh, Kamuya, & Molyneux, 2011). As a person's sense of guilt in such circumstances is often the result of complex interaction between self-assessment, beliefs about causality and the responses of others (Weil, 2000), a general attitude of blame towards mothers, such as described by Marsh *et al.* (2011) could make those mothers particularly vulnerable to depression or other mood disturbances.

Parents or caregivers of children with SCD face a variety of potentially problematic situations relating to raising an affected child. These situations can be illness specific, and as such different from general child rearing problems faced by all parents. It can include ensuring good nutrition, minimizing pain episodes, managing medication and helping the child cope with negative feelings about having SCD, academic problems and social issues (Ievers-Landis *et al.*, 2001).

With regards to nutrition, research has shown that ensuring a child affected with SCD avoids dehydration and exhaustion is a problem that arises regularly (Hyacinth, Adeyeke, & Yilgwan, 2013; Ievers-Landis *et al.*, 2001). Although any parent would be reasonably worried about preventing these situations, parents of children with SCD are often aware that in doing so they could also be preventing a vasoocclusive or pain crisis. As such, these parental tasks can become much more time consuming and stressful, since the potential risks for significant morbidities are much greater than that of the average child.

The time needed to care for a child with a chronic disease, such as SCD, can be challenging, as it includes both technical and non-technical care. An example of technical care would be the administration of medication, while non-technical care includes feeding, grooming and dressing (Moskowitz *et al.*, 2007). Moskowitz *et al.* (2007) found that the care demands of children with SCD are such that approximately 1.5 hours per day is spent on illness related care alone. This is over and above time spent caring for a healthy child of similar age. This level of time commitment can also have additional consequences, impacting other parental responsibilities, such as care of other children, employment, household management and social activities (Moskowitz *et al.*, 2007).

Financial concerns are often associated with increased parental stress. As expected, this is also true in the case of parents of a child with SCD, and a Nigerian study by Brown *et al.* (2010) reported a significant correlation between stress, finances, and parental difficulty in dealing with episodes of SCD illness. The study further reported that reduced employability of the carer has been associated with caring for a child with SCD (Brown *et al.*, 2010). This is doubtless as a

result of absences due to frequent hospital visits and the unpredictability of pain crisis, which can interfere with work commitments and have been described in a few studies (Moskowitz *et al.*, 2007; van den Tweel *et al.*, 2008; Wonkam, Zameyo Mba *et al.*, 2014).

There are a number of expenses related to caring for a child with SCD, of which the cost of treatment is undoubtedly the greatest. This particular financial burden can be significantly reduced by accessing government health insurance, which offers state subsidies to lighten parents' financial responsibility. This service is, however, not available in all countries, including several on the African continent, or does not necessarily include SCD treatment. Fullwiley (2010) describes SCD financing difficulties in Senegal as a 'state neglect of widespread diseases like sickle cell'. As a result of economic triage in that country, majority of available funds go to global health priorities, such as HIV/AIDS and malaria, and cost of SCD treatment is the responsibility of the family (Fullwiley, 2010).

Apart from treatment, there are also additional costs involved in caring for a child affected with SCD, such as travelling to the hospital and loss of income through care related absences. Health status of a child with SCD can have a considerable impact on the socioeconomic burden placed on families (Wonkam, Zameyo Mba *et al.*, 2014).

Coping with pain is often reported by parents to be the most trying situation arising from the disease (Ievers-Landis *et al.*, 2001; Moskowitz *et al.*, 2007). Pain crises in SCD can occur unexpectedly, and are therefore difficult to prepare and provide care for, often leaving parents with feelings of inadequacy and guilt. It is not surprising that Moskowitz *et al.* (2007) found caregiving burden to be directly associated with the amount of crisis care needed by the affected child. Similarly, a study done in Cameroon by Wonkam *et al.* (2014) found that pain crises most undermined the coping ability of parents. These pain crises can cause further parental anxiety by resulting in school absenteeism and contributing to poor academic achievement (Brown *et al.*, 2010; Wonkam, Zameyo Mba *et al.*, 2014).

The chronic nature of SCD may also contribute substantially to reduce social functioning, quality of life as well as alter the inter- and intra-personal relationships of patients (Brown *et al.*, 2010). Parents of affected children often report daily emotional challenges, including constant fear of hospitalisation, possible death, separation anxiety and feelings of helplessness and loss of control.

Promoting the wellbeing of caregivers is important as this can often indirectly influence the quality of care given to the affected child (van den Tweel *et al.*, 2008).

1.2.5 Screening for Sickle Cell Disease

Screening can be defined as the systematic application of a test to identify individuals at sufficient risk of a specific disorder to warrant further investigation or direct preventative action, amongst persons who have not sought medical attention on account of symptoms of that disorder (Davies *et al.*, 2000). And while screening for SCD is encompassed in that definition, as a genetic condition, there are also other important points to consider. Firstly, genetic screening should contribute to the health of the person suffering from the condition, and not merely reveal risk. Secondly, it should allow carriers to make informed choices regarding reproduction and lastly, genetic screening should move towards alleviating the anxieties of families who are faced with the prospect of serious genetic disease (Davies *et al.*, 2000).

When implemented correctly and successfully, screening for SCD covers all the above mentioned points; can lead to increased awareness and education of SCD (Carey, 2014); and, most importantly, lead to a reduction in disease incidence (Davies *et al.*, 2000). Screening for SCD is most often done at three stages: premarital, neonatal and prenatal testing.

Premarital Screening

Premarital genetic screening involves the detection of SCD mutation carriers and informing them of their risk of having an affected child prior to their marriage and/or childbearing years. For autosomal recessively inherited diseases, such as SCD, this can possibly be described as the only available form of primary prevention, as prenatal and neonatal screening are both secondary preventions (Memish & Saeedi, 2011). Carriers are accurately and inexpensively detected through routine haematological methods and can then be forewarned of their risk, which allows couples to make reproductive choices. These factors make premarital screening an important strategy to reduce disease incidence and policies implementing it can lead to a fall in SCD birth incidence (Modell & Darlison, 2008).

A recent study in Saudi Arabia reviewed the impact of five years mandatory premarital screening on the prevalence of haemoglobinopathies. While no significant decrease in SCD prevalence was noted as yet, the study did find a 60% decrease in at-risk couples over the five

year period, which may considerably reduce the disease burden over coming decades (Memish & Saeedi, 2011). Similarly, through the prevention of at-risk marriages and education on options such as medical abortion, premarital screening has proved successful in a number of countries including the USA, UK and Canada (Alswaidi & O'Brien, 2011).

Neonatal Screening

The principal aim of neonatal screening is to identify newborns with SCD, which allows for certain early interventions such as antibiotic prophylaxis and good nutrition (Carey, 2014). The USA started its first newborn screening program for SCD in 1975 (Ashley-Koch *et al.*, 2000) based on the belief that early diagnosis would improve morbidity and mortality. The belief proved to be true, with early studies reporting a decrease in infant mortality of 10% (Vichinsky, Hurst, Earles, Kleman, & Lubin, 1988). In recent years SCD mortality in the USA has decreased drastically, primarily because of early intervention, better medical care and education of family members (Ashley-Koch *et al.*, 2000). Due to the success of these genetic screening programs, many other countries have started implementing similar programs, in the hopes of improving health care and outcomes of affected individuals (Makani *et al.*, 2013).

Although not much research has been conducted in Africa on approaches to and views on neonatal genetic screening for SCD, Marsh *et al.* (2013) in Kenya identified a few difficulties that might arise due to SCD screening. These difficulties included parental denial, paternal denial through doubts around paternity, managing incidental findings on non-paternity and maternal blame (Marsh *et al.*, 2011; Marsh, Kombe, Fitzpatrick, Molyneux, & Parker, 2013). To overcome these problems, it is important to combine neonatal screening with genetic counselling and disease education (Modell & Darlison, 2008).

Indeed, to maximise the benefit of neonatal screening, a positive SCD test result should be accompanied by enrolment into a health service that provides comprehensive care by a multidisciplinary team (Makani *et al.*, 2013). The team should be comprised of genetic counsellors, nurses, social workers, paediatricians, haematologists, orthopaedic surgeons and internists. Appropriate advice, counselling, support and care can then be provided to parents and affected individuals throughout the course of the illness.

Prenatal Testing

One particular challenge for couples who already have a child affected with SCD is the possibility that future children may also be affected, adding to the financial and emotional burdens already experienced by the family. A possible solution to this is prenatal genetic diagnosis (PND), which affords the parents the knowledge of whether the foetus is affected before the child is born. PND represents a type of preventative strategy, as it would allow parents the possibility of terminating the pregnancy or taking early precautions against secondary complications. This can also be done for first time parents, following positive premarital screening. Genetic counsellors play an integral role in creating awareness of PND and the availability, optimal timing, and utility thereof.

The acceptability of prenatal testing and subsequent diagnosis and termination varies. Little is known about the attitudes toward PND and termination of pregnancy among parents of SCD affected children in sub-Saharan Africa (Wonkam *et al.*, 2011), and while the option has been available in Cape Town since 2010, even less is known about the attitude of parents in SA. Studies have shown that parents of affected children in Cameroon show a surprisingly high acceptance of termination of a SCD affected foetus (Wonkam, de Vries, Royal, Ramesar, & Angwafo, 2014; Wonkam *et al.*, 2011). It is unknown if this is also the case among parents in SA. Preliminary data from an investigation done in 2012 on the awareness and attitudes of parents with SCD affected children in Cape Town showed that the majority of parents (70%) were unfamiliar with PND. It concluded that the majority of people would accept the option of PND if they were to fall pregnant again, but the possibility of termination was rejected by most (66.6%) (Wonkam *et al.*, unpublished data).

The question of PND warrants further investigation, as the uptake thereof could lead to a reduction in mortality rate due to early intervention. Consequently, it is necessary to determine the knowledge and awareness of PND in Cape Town in order to plan appropriate preventative genetic services.

Feelings on all the above mentioned preventative strategies often vary with social attitudes and can be more or less effective, depending on the religious, cultural and moral beliefs of the population (Modell & Darlison, 2008). The effect and success of screening relies on the choices made by the concerned individuals (Davies *et al.*, 2000). Premarital testing allows for the widest range of choices while requiring the least amount of laboratory tests. In contrast, a prenatal test

gives fewer options and requires more invasive testing (Modell & Darlison, 2008). The success of each strategy might depend on how comfortable individuals are with the choices they provide. For this reason, it is imperative that screening services not only provide testing and consequent options, but also an explanation of all relevant information and guidance in the process of making an informed decision.

1.2.6 Sickle Cell Disease in South Africa and Cape Town

South Africa, being an area with little to no malaria exposure, originally had a low incidence of SCD, although prevalence differs for each ethnic group (Wonkam *et al.*, 2012). Literature on SCD prevalence in the local South African populations is rare, but it has been found that in the mixed ancestry population of SA, several haemoglobin variants occur, HbS and HbE being the most common occurring at a prevalence of 1% each (Bird, Ellis, Wood, Mathew, & Karabus, 1987). In addition to the mixed ancestry population, SCD cases have also been described in the South African Indian population as well as South African Caucasians not of Mediterranean descent (Wonkam *et al.*, 2012). With regards to the prevalence of SCD among the African populations of SA, the heterozygous trait can be found among the indigenous Venda and Shangaan ethnic groups at a frequency of approximately 0.2% (Wonkam *et al.*, 2012). This low frequency, along with the fact that the ethnic groups mentioned are two of the least numerous in SA, makes SCD very uncommon among indigenous African people from SA.

The demographics of SA are changing, however, due to a redistribution of migrants from other parts of Africa, particularly Central Africa, where SCD is highly prevalent (Wonkam *et al.*, 2012). Wonkam *et al.* (2012) found that up to 93.1% of SCD patients seen in Cape Town paediatric hospital were originally from other African countries. With this influx of immigrants, there now exists in SA a thriving cohort of adults and children affected with this previously infrequent disease. In Cape Town alone, the inflow of immigrants has led to an increase in SCD frequency of up to 400% over the decade ranging from 2001 to 2010 (Wonkam *et al.*, 2012).

This increase necessitated an adjustment of services offered to allow for the effective treatment of all affected individuals. Red Cross War Memorial Children's Hospital (RCWMCH) is a tertiary paediatric hospital where the Haematology/Oncology Service has developed SCD-specific treatment guidelines and since 2009 sponsored the development of satellite clinics at New Somerset Hospital and Victoria Hospital, two secondary-level hospitals in the Cape Town metropolitan area. This adjustment is still ongoing. One suggestion made to improve care of

SCD patients and caregivers is that genetic counselling become an intrinsic part of their health care (Modell & Darlison, 2008), and the WHO lists genetic counselling as a priority intervention in primary prevention of SCD (World Health Organization, 2010). While genetic counselling services are available at RCWMCH, not all patients are referred to or make use of this service and genetic counselling services do not extend to surrounding public primary or secondary level hospitals.

1.3 Rationale for study

As discussed in the literature review, parents and caregivers of children with SCD face many psychosocial burdens, including financial, emotional and disease related. There is a resident Cape Town population of SCD patients, but no understanding about the burdens they face or their experiences in having an affected child. It is also unclear what is known about SCD in this population, and how that knowledge might influence their experience.

Further exploration into the role of genetic counselling in the care of SCD patients and caregivers in Cape Town is important, as this service could play an invaluable role in helping parents understand and cope with the initial diagnosis, as well as being a continued part of the support structure available to the patient and family.

1.3.1 Aims

In order to better attune current genetic counselling services offered to caregivers, this study aims to explore the knowledge and understanding of SCD among parents of SCD affected children in Cape Town and identify burdens associated with caring for an affected child. Furthermore, this study aims to discover parents' attitudes toward certain preventative policies and assess opportunities of improving the genetic counselling services available to caregivers in Cape Town.

1.3.2 Objectives

- To conduct 15-20 interviews with suitable participants.
- To investigate the participants' knowledge of SCD.
- To identify factors contributing to burden's experienced by participants.

- To investigate participants' feelings on preventative policies for SCD.

The following chapter describes the methodological approach used to investigate and explore the aims and achieve the objectives.

Chapter 2: Methodology

2.1 Chapter introduction

The previous chapter outlined the literature on the subject of SCD that served as a background to this study. It described how genetic counselling services could potentially play a vital role in improving care for individuals affected with SCD or helping caregivers manage the psychosocial burden better. This chapter describes the methodological framework used for this study, the population and sample used, participant recruitment, measurement instruments and how academic rigour was maintained. It will also provide an explanation of data collection and analysis process.

2.2 Research design

This study was conducted using qualitative research methodology. Qualitative studies are a form of research characterized by a drive towards in-depth understanding of a certain topic or circumstance, as opposed to being driven by the aim to generate accurate predictions (Patton & Cochran, 2002). Qualitative research involves an interpretive, naturalistic approach to the world, which means that the researcher studies things in their natural settings, attempting to make sense of phenomena in terms of the meanings people bring to them (Creswell, 2013). The understanding sought is acquired through the narration of the participants' meanings for these phenomena. The meanings can include the participants' feelings, ideas, beliefs, thoughts and actions (McMillan & Schumacher, 2001). One of the strengths of qualitative research is that it draws on participants' individual life experiences and the context within which they live.

Qualitative research is conducted when a researcher wants to gather an in-depth understanding of a research question (Creswell, 2013). This can be the case when literature on the topic is limited, or, as is the case with this study, because existing literature does not take into account certain characteristics of the population being studied. Moreover, certain research questions lend themselves better to qualitative research, as they need to draw on people's individual experience to form an answer. As this research aimed to explore personal experiences of SCD in a population hitherto unexplored a phenomenological, cross sectional approach was used.

Phenomenology is an approach to qualitative research that describes the common meaning different individuals attribute to their experiences of a phenomenon (Creswell, 2013). It focuses on what all participants have in common as they experience a phenomenon. The study

investigated participants at a single point in time, which required the use of a cross sectional design. In comparison, a longitudinal study would investigate the same participants over a period of time. While a cross sectional study has the disadvantage of not taking into account any data that might change over the course of time, it has the advantage of being less time consuming. The latter made it an ideal approach for a minor dissertation.

2.3 Study population and sample

2.3.1 Population

The majority of diagnosed SCD patients enrolled for healthcare in Cape Town are children, most of whom either immigrated to SA with their parents, or are first generation South Africans. Due to their age, genetic counselling services would not focus on these patients themselves, but rather on their parents or primary caregivers. For this reason, this study enrolled all consenting parents that presented at RCWMCH Haematology Clinic with their SCD affected child in February and March of 2015.

Only biological parents were recruited, as this was the most time effective way to discover the primary caregivers most likely to have the in-depth experiences sought in this study. Furthermore, only parents who could speak English were recruited. This was done as the time constraints of a minor dissertation made the use of various translators impractical. However, a recent review of parents of SCD patients attending RCH reported only 3 out of 56 (5.3%) as unable to speak English. Therefore it was felt that this exclusion was unlikely to eliminate large numbers of possible participants. The study recruited parents of both genders, as the experiences of a father might be different to those of a mother. However, the recruitment strategy was to interview whoever brought the child to clinic, which, in most cases, proved to be the child's mother. This is reflected in the final sample demographics as described in Chapter Three.

2.3.2 Sample size

The intent in qualitative research is not to generalize the information, but rather to elucidate the specific (Creswell, 2013), and given that the purpose is to acquire in-depth and multi-dimensional data, a small, information-rich sample is sufficient (B. Marshall, Cardon, Poddar, & Fontenot, 2013; M. N. Marshall, 1996). Prior to starting the interviews, and based on precedents by similar studies, it was estimated that a total of 15-20 interviews would be conducted, continuing until saturation was reached. Keeping in mind these considerations, the researcher

recruited 17 participants, 12 female and 5 male. The nature of a minor dissertation and time constraints prevented the recruitment of a larger sample size. However, it was felt that data saturation had been reached.

2.3.3 Sampling method and recruitment

Participants were selected using both purposive and convenience sampling methods. Purposive sampling involves recruiting participants due to certain qualities they possess and specific criteria that they meet (Treadwell *et al.*, 2006), while convenience sampling involves drawing samples that are both easily accessible and willing to participate in a study (Teddlie & Yu, 2007).

The researcher attended Haematology clinics at RCH, where she was already known to the nurses and doctors, having previously completed 3 months clinical training there. Two members of the supervisory team are also involved in this clinic as medical professionals.

The nurse running the clinic initially pointed out any eligible participants, i.e. parents who had brought their affected child to clinic. The researcher subsequently approached possible participants to inform them of the objectives and methods of the proposed study. Prospective participants were given an information sheet (Appendix I) to read. The researcher also used this initial approach to establish their proficiency in English, thereby excluding those who did not meet that particular inclusion criterion.

Individuals willing to participate were interviewed at a convenient time, usually during clinic hours on the same day, but on one occasion by appointment at a later date. Participant recruitment and interviewing took place between February and March 2015.

2.4 Measurement instruments

For the purpose of this research, a semi structured interview guide was used as an instrument to assist in data collection (Appendix II). The guide covered several topical areas, and enabled the researcher to explore participants' thoughts, feelings and opinions without the fear of anything significant being overlooked. This type of interview does not require questions to be asked in a particular order, but allows the flexibility for any emerging topic of interest to be discussed as and when it arises from the dialogue between interviewer and interviewee (Patton & Cochran, 2002).

The interview guide was developed by identifying potentially relevant themes through the literature review, and by examining previous guides used in similar research in other settings. The interview guide identified important themes for investigation, as well as a number of sample questions for each theme. In addition to open-ended questions, basic socio-demographic information was gathered at the start of each interview. The use of open-ended questions allowed and encouraged elaboration on the part of the participants, giving them the chance to express themselves freely and in great depth on those topics they considered most important. The interview guide included questions in the form of key words or phrases that served as a prompt to the researcher. These words or phrases could then be adapted into open-ended questions as needed. The final interview guide consisted of 24 prompts in total, divided into four sections: sociodemographic information, knowledge and understanding of SCD, experiences with SCD and preventative policies.

In order to ensure validity of the guide the following procedure was followed in developing it: the first draft of the guide was critically reviewed by the supervisory team to ensure that all relevant questions were included. The guide was then used as a base for the first pilot interview, which was a role play scenario involving the student researcher and the genetic counsellor on the supervisory team. A second genetic counsellor and trained qualitative methodologist observed the interview. Following this first pilot, the interview guide was tested in a second pilot interview with a participant who met the inclusion criteria. Feedback and lessons learnt from these two pilot interviews were incorporated in the guide. It was then reviewed finally by the supervisory team and used for all subsequent interviews. As the participant of the second pilot interview met all the inclusion criteria, she was ultimately included in the study subsequent to a follow-up interview to augment the data already gathered.

2.5 Data collection

2.5.1 Research setting

Data collection took place at the Haematology clinic at RCWMCH, in what can be called the 'natural setting', or the site where participants experience the problem being studied (Creswell, 2013). In this case the 'natural setting' was the setting in which they were faced with the medical aspects of SCD. This setting was convenient as it involved no extra travel or cost to the participants and interviews could often be done during time that would have been spent in the waiting room. While interviewing participants in their own home would have given the

researcher the opportunity to observe them within their home context, the logistical difficulties of travelling and arranging suitable meeting times made this option impractical within the available time.

As mentioned, the researcher had previously spent time working in the Haematology clinic. This time spent in the environment was invaluable as it gave the researcher some time to observe the setting, possible future participants, and orientate herself in the field where data collection would take place. Interviews were conducted in a semi-private room due to the impracticality of ensuring a completely private room at all times. However, brief interruptions, when they occurred, were by clinic staff well known to the participants, and did not seem to impede their willingness to discuss sensitive topics in any way.

2.5.2 Procedure

There are several methods of collecting data in a phenomenological approach, from individual interviews to diary entries. In this study, data needed to answer the research questions was gathered through semi-structured interviews held with research participants. Face-to-face interviews are one of the most common approaches to the collection of qualitative data, as it allows for the detailed exploration of any personal and social matters that would not be easily discussed in a group setting (Dicicco-bloom & Crabtree, 2006; Sturges & Hanrahan, 2004). The fact that the interviews were done in person allowed the researcher to observe and note any nonverbal communication or behaviour that possibly added to the richness of the data. It also enabled the researcher to react to any cues from the participants in order to achieve maximum clarity on the information shared.

All interviews were conducted by the researcher and lasted on average 40 minutes. Interviews were audio recorded in order to avoid excessive note taking during interviews. This allowed undisturbed observation and engagement between the researcher and participant. Furthermore recording the actual words spoken by participants allowed for re-examination of interviews during transcription.

While recordings were the main source of data, there were multiple secondary forms of data in the shape of descriptive notes taken during the interview and directly afterwards as well as reflective notes developed over time while studying the data. Together, these notes are referred to as field notes in the remainder of this work.

Participants were interviewed over a period of eight weeks. Before the start of every interview, the researcher and participant went through the informed consent form (Appendix III), discussing the study objectives and matters of confidentiality. Participants were reassured that anonymity would be maintained through the use of unique identifiers assigned to each interview. This would be the only form of identification used in the interview guide, transcript and data in the dissertation. Personal details of the interviewees were not shared with the supervisors.

Discussion of the consent form together with the initial demographic questions asked was a good opportunity for the researcher to establish a rapport with the participant, making it easier to ask more personal questions as the interview progressed. Following the interview, participants were referred to a qualified genetic counsellor when necessary or if they expressed a wish for further information regarding the condition.

2.6 Data analysis

Data generated during the interview process are so-called raw data that have no meaning on their own unless processed, organised and interpreted to identify important findings. This study made use of thematic content analysis to explore the data and identify underlying themes (Creswell, 2013). This method of data analysis was selected as it focuses on the extraction of "content areas" or "coding" so as to categorize and analyse themes to create a comprehensive picture of an individual's experience of a phenomenon.

In order to prepare the data for analysis, all interviews were transcribed by the researcher. This happened concurrently and subsequent to the interviews. Engagement in transcribing was an opportunity for the researcher to become familiar with the data, getting an idea of each interview as a whole before breaking it into parts through analysis. Listening to the interviews while transcribing also helped the researcher acquaint herself with the participants, their way of speaking and their vocabulary. All this became valuable in the form of reflective field notes to inform data analysis and conclusions.

NVivo software (QSR International, 2012) was used to assist in the analysis and coding of transcripts. Coding the data refers to the process of reducing it into meaningful segments and assigning names to those segments (Creswell, 2013). This process of interpreting qualitative data can be differentiated further into open coding, axial coding, and selective coding (Flick, 2009).

These procedures can be combined or moved between in order to manage data. The process begins with open coding.

In the process of open coding, the researcher explores broad themes in two indicator interviews. By reading through two transcripts and open coding them, a list of tentative codes was developed. Certain codes overlapped between the two transcripts, while new ones were created whenever needed to match text segments. In this manner a preliminary list, consisting of more than 50 codes, was created. The researcher met with members of the supervisory team, who had independently coded the same two transcripts. Following this meeting, the team agreed to code a further three interviews before a good coding matrix could be arrived at. These further transcripts were used to eliminate redundant codes, develop ideas about code hierarchies and identify emergent categories. Data from all five transcripts were then organized inductively into increasingly more abstract and hierarchical codes. Patterns, categories and themes were built from the bottom up, forming hierarchies based on relationships and combining them into broader categories or themes.

Themes in qualitative research are broad units of information consisting of several codes aggregated to form a common idea (Creswell, 2013). Following this process, another supervisory meeting was held where the coding scheme was discussed and finalised. The resulting hierarchical coding matrix was then applied to all transcripts, including the five that were coded for the development of the open coding scheme. In analysing all the transcripts, certain codes became less important, while others emerged even more strongly as themes.

Once all the transcripts had been analysed, each code and all the references for that code were printed to facilitate analysis. The researcher then studied the references, group by group, concurrently looking through field notes made during interviews and transcription in order to start forming theories about the data and how it informed the aims of the study. Creating links and relationships between various concepts and references, the researcher looked at how the data interacted and influenced one another, trying to find the larger meaning of the data. From there the results and discussion were written up.

2.7 Validity and reliability

Validity is traditionally defined as a determination of whether research truly measures what it was intended to measure. In qualitative research, validity can be seen as an attempt to assess the

accuracy of the findings, as described by the researcher and participants (Creswell, 2013). However, a basic problem in assessing the validity of qualitative research is how to specify the link between the situations that are studied and the version of them presented by the researcher. In this context, a position of "subtle realism" can be outlined, starting from three premises (Flick, 2009). Firstly, the validity of knowledge cannot be assessed with certainty and assumptions should be judged based on their plausibility and credibility. Secondly, phenomena also exist independently of any claims made concerning them. Any assumptions made can only more or less approximate these phenomena. Lastly, reality becomes accessible across the different perspectives on phenomena and research aims at presenting reality, not reproducing it (Flick, 2009). Starting from this position, validity in qualitative research becomes about how far the researchers' constructions (i.e., perceptions, interpretations, and presentations) are grounded in the constructions of those whom they studied, as well as how far this grounding is transparent to others (Flick, 2009). Thus, the production of data can be used to assess validity together with the presentation of phenomena and the inferences drawn from them.

To increase validity in the present study, the researcher regularly met with her supervisory team who, through peer review and debriefing, provided an external check for the research process (Creswell, 2013). All methods used, as well as emergent themes, were carefully documented, analysed and discussed so as to ensure academic rigour and transparency.

Reliability refers to the constancy or consistency of results obtained (Flick, 2009) and requires a detailed account of the decision making process so others may follow the same procedures. In order to ensure consistent results, all interviews were conducted and analysed by the same person.

2.8 Ethical considerations

2.8.1 Ethical approval

Ethical approval for this study was granted by the Faculty of Health Sciences Health Research Ethics Committee of the University of Cape Town (FHS HREC/REF: 905/2014, Appendix IV).

2.8.2 Consent

Individuals meeting the inclusion criteria were approached by the researcher to explain the purpose of the study and gauge their interest in participating. Assurance was given that all

information given in the course of the interview would be kept confidential. Data would be anonymised and destroyed upon completion of the research. It was stressed that participation is entirely voluntary and would not result in any medical or financial gain. Participants had the right to withdraw from the study at any stage should they so wish, without incurring any (medical or other) repercussions.

Prior to the start of each interview written informed consent was obtained from each participant. This included consent for the interview to be audio recorded.

2.8.3 Confidentiality

Confidentiality and anonymity were a priority in this study and maintained by the researcher throughout the research project. Both the recordings and subsequent transcripts were kept on a password protected computer, to which the researcher alone had access. These records will be destroyed once the research has been published. To ensure anonymity no names were used in transcripts, with all participants instead being assigned an identifier. These identifiers were used throughout when referring to a specific participant. In transcripts and quotations, anonymity was further sustained by referring to people mentioned by the participant by their relationship to that participant (her daughter, his son) instead of using names.

Personal details of interviewees were not shared with the supervisory team unless the participant requested follow-up with a qualified genetic counsellor (who was part of the supervisory team). In that case, certain personal details were shared as well as the reason for the request for counselling. However, entire interview transcripts were not shared and the qualified counsellor did not at any time know the specific identifier assigned the individuals referred for genetic counselling.

2.8.4 Risk and benefit to participants

Risk to the participants included the discussion of sensitive and emotional topics. The researcher tried to be sensitive to the emotional state of the participants, ensuring any actionable comments were acted upon, and referring participants to a qualified genetic counsellor if needed. Despite being an emotional experience, there can be a therapeutic benefit to discussing one's understandings and experiences which may have been experienced by the participants. They also benefited from the opportunity to ask questions and the provision of contact details for further information. The long term benefit of the study is to employ the research findings to, if needed,

improve the genetic counselling process and services provided to parents of children affected with SCD in Cape Town.

2.9 Chapter summary

This research project focuses on parents of children affected with SCD in Cape Town and exploring their personal experiences and knowledge around their child's diagnosis. This chapter described the methodology used to generate empirical data that would help achieve the aims of this study, as discussed in Chapter One. A total of 17 qualitative interviews were conducted with parents of children with SCD living in Cape Town. Interview data were analysed thematically to identify the main themes emerging from the participants' discussion. The next three chapters will describe and discuss the results of this analysis, divided by subject into: knowledge; experiences; and screening and genetic counselling.

Chapter 3: Results and Discussion on Knowledge of Sickle Cell Disease

3.1 Chapter introduction

This chapter starts with a brief description of the study participants, followed by the findings and discussion of their knowledge of SCD. This includes whatever participants knew about SCD prior to their child's diagnosis and the different ways in which this knowledge can influence their current view of the illness. Furthermore, participants' cognisance and understanding of the cause and inheritance pattern of SCD will be discussed, together with any assumptions or misconceptions that arose from this knowledge. An understanding of these misconceptions is important as it can help improve genetic counselling services by revealing areas of confusion.

3.2 Sociodemographic data of participants

All participants but one had immigrated to SA from other African countries. Majority of participants (n=10) were originally from the Democratic Republic of Congo (DRC). Age of participants ranged from 30 - 50 years and most were married. All but one of the participants described themselves as religious, with the majority being Christians (n=14), with two Muslim participants and one Jehovah's Witness (Table 1).

The number of children in a participant's family ranged from one to five, with the average being three. Most participants (n=15) had only one confirmed SCD affected living child, but two participants had two affected children each. One participant was awaiting SCD test results on a sibling of their affected child, while another had a previous child who she now suspected had died of SCD related complications. All participants' children were affected with SCA, the most common form of SCD.

The majority of participants completed high school, and some had gone on to complete tertiary education (Table 1). Participant P12F was completing a nursing degree - the only the participant with any specialized education in the field of health care. Two participants had attended, but not completed high school, and a further two attended only primary school. A summary of the sociodemographic data of the participants is presented in Table 1.

Table 1: Summary of participants' sociodemographic information.

P	Age	Marital status	Religious Affiliation	Level of education	Country of origin (participant)	Country of origin (partner)	Number of children	Number of affected children	Age of affected children
P1F	39	Married	Christian	Secondary	DRC	DRC	4	1	14
P2F	44	Single	None	Secondary	RSA	Nigeria	2	1	10
P3F	30	Married	Christian	Tertiary	DRC	DRC	2	2	8 and 2
P4M	34	Married	Muslim	Secondary	DRC	DRC	3	2	8 and 4
P5F	30	Married	Christian	Secondary	DRC	DRC	3	1	2
P6M	30	Married	Christian	Secondary	ZIM	ZIM	1	1	5
P7M	30	Married	Christian	Secondary	DRC	DRC	3	1	5
P8F	42	Married	Christian	Secondary	DRC	DRC	2	1	13
P9M	43	Married	Christian	Secondary	DRC	DRC	5	1	4
P10F	36	Married	Muslim	Primary	Malawi	Malawi	3	1	11
P11M	50	Married	Christian	Secondary	ZIM	ZIM	3	1	13
P12F	37	Married	Christian	Tertiary	ZIM	ZIM	2	1	7

P13F	47	Married	Christian	Tertiary	Uganda	Uganda	3	1	14
P14F	38	Married	Jehovah	Primary	DRC	DRC	3	1	9
P15F	29	Single	Christian	Tertiary	Gabon	Gabon	1	1	5
P16F	36	Married	Christian	Tertiary	DRC	DRC	3	1	9
P17F	42	Married	Christian	Secondary	DRC	DRC	3	1	7

P: Participant; M: Male; F: Female; DRC: Democratic Republic of Congo; RSA: Republic of South Africa; ZIM: Zimbabwe

3.3 Parents' knowledge of Sickle Cell Disease

3.3.1 Introduction

In an illness like SCD, parental understanding of the disease and its causes has both physical and psychological value. At a physical level, knowledge can be crucial for optimal self-management of the disease in order to reduce long term complications, while psychologically it can help improve psychosocial adjustment and adaptive coping (Bhatt, Reid, Lewis, & Asnani, 2011).

This psychological benefit is arguably the more valuable of the two. Various studies have described a sense of security that comes from knowledge about a genetic diagnosis and it is often found to be a way of gaining control of an otherwise ambiguous situation (Long, Thomas, Grubs, Gettig, & Krishnamurti, 2011; Skirton, 2001).

But, while knowledge can be beneficial, it can be equally detrimental, especially when the information is incorrect. A few studies have described important misconceptions that exist about SCD (Acharya, Walsh Lang, & Friedman Ross, 2009; Long *et al.*, 2011), and to a certain extent those findings were echoed in this study.

3.3.2 Disease name

All participants were asked if they had heard about SCD before their child was diagnosed. This was asked in order to evaluate their prior knowledge of SCD. Several participants responded that they had never heard of SCD before, not because they were unfamiliar with the disease, but because they knew it under a different name. The term Sickle Cell Disease was often one they only became acquainted with once they started attending clinics in SA. Marsh *et al* (2011) reported a similar unfamiliarity with the term among Kenyan families, who referred to SCD instead by certain names in their native languages.

In this study, participants from DRC were generally more acquainted with the name “SS”. This name is a reflection of the kind of haemoglobin found in a person affected with SCA, the most common form of SCD. As mentioned in the literature review, individuals affected with SCA will have two copies of the variant HbS, commonly referred to as ‘SS’. It has become the practice of health care workers to refer to the haemoglobin simply as ‘SS’, or ‘CS’ (if affected with Sickle Haemoglobin C disease), and in many regions it appears as if this name has become synonymous with the disease, possibly stemming from patients’ familiarity with ‘SS’, ‘AS’ an ‘AA’ as terms to describe affected, carrier and unaffected individuals respectively. This concept is illustrated by the following excerpt from an interview:

P3F: “We just say SS.”

Interviewer: “And then that means the whole disease ... all the kinds of Sickle Cell?”

P3F: “Yes, you see, I’ll say she is SS, or she’s got SS.”

The use of the term ‘SS’ as a name for SCD can lead to a number of misunderstandings, which will be discussed in section 3.3.5.

Two participants, P4M and P9M, referred to the disease simply as “*anémie*”, the French word for anaemia. Both these participants were from DRC, where the official language is French. The name refers to one of the main symptoms of SCD, but as used by these participants, there appeared to be no way of distinguishing it from other forms of anaemia. It is unclear whether in using this name, the participants were referring only to SCA or if the name would apply equally to all types of SCD.

Two of the participants from Zimbabwe (P6M and P11M) also mentioned SCD as having different names in their community, “Sickler” and “Yellow”. The term ‘sickler’ as a label for someone affected with SCD has often been experienced as offensive by patients as it reduces them to nothing more than their disease (Alleyne & Thomas, 1994). However, participant P6M did not show any noticeable feelings of stigma relating to the name, even upon further probing, and it is possible that in certain areas the word has no negative connotation. Participant P11M said that where he came from, the disease was simply called “Yellow”, after the haemolysis related jaundice that often lead to the yellow colour of the sclera of a SCD-affected person.

3.3.3 Prior knowledge and its influence on present perception

Only one participant had never heard of SCD, under any name, before her child’s diagnosis. The particular participant, P2F, is a South African where, as mentioned, SCD is not very common and thus fairly unknown among the general population. Another participant had no personal knowledge of SCD, but had come across it in her tertiary education. The remaining participants all had varying degrees of knowledge about SCD even before their children had been diagnosed, either through a family or community member who was affected or, in the case of certain participants from DRC, because of the high prevalence of the disease in that country.

While discussing this prior knowledge of SCD, it became apparent that all participants who had some prior knowledge of SCD had consequently developed a very negative view of what the disease was and what the outcome would be in terms of progression and life expectancy. For example, several participants shared their views about the characteristic appearance of a child with SCA in parts of Africa:

P3F: “Just by looking at them you can tell that this one is a Sickler Cell, you don’t even have to wait for someone to tell you. They look sick.”

As is illustrated by the above comment, it seems that many participants felt that SCA could be identified by a certain ‘look’ that children affected therewith had. Specifically they described the yellow eyes, distended stomach and pale skin that can characteristically affect a child with SCA. One participant likened the appearance to that of someone with Kwashiorkor, a form of malnutrition typically affecting children in tropical areas.

In certain cases, this ‘look’ seems to have become almost synonymous with a diagnosis of SCA, and as such, it is assumed children who do not look this way cannot have SCD. This can lead to considerable confusion for some parents, as the absence of these distinctive characteristics can lead to doubt when a diagnosis is given. The belief in this physical display of SCD due to prior knowledge was so pronounced that several participants mentioned not believing the diagnosis as their affected children did not have the look they expected:

P3F: “She (neighbour) was looking different from other people. When you see her she was having yellow eyes, she was having wounds every time...”

It’s difficult for me to accept.”

Interviewer: “Why is it difficult to accept?”

P3F: “Because of those I have seen before.”

The second main point arising from discussions on prior knowledge on SCD was participants’ expectations on disease progression. Most of the participants are from countries where treatment is scarce and expensive, and as a result the prognosis of SCD in these countries is not as good as in more developed countries, such as the USA and UK (Modell & Darlison, 2008; Weatherall & Clegg, 2001).

While it is true that SCD is a chronic illness, it was also clear that most participants were describing an expectation of something much worse than what is typically seen in children with SCD in SA receiving adequate treatment. This perception among participants had been formed by experiences, either personal or indirect, of SCD in their country of origin.

For instance, it was the expectation of several participants that someone affected with SCD would have a strikingly shortened life. Participants P10F and P17F used the word ‘lucky’ to describe children with SCD that survived to 5 and 10 years respectively. Participant P5F had the following to say on life expectancy:

“For me, it’s like a big disease and in Congo, people with this kind of disease is a huge problem. They’ve got limited of life. In Congo, this disease is expensive to treat, so most of the

people used to die at the age of twenty years. They don't get to twenty-five years."

The idea of affected persons only surviving until the age of 25 was echoed by another participant, P9M, who said:

"My son, because he is category of "anémie", he not get age of 25, because that is the experience I get from my country. Those that have "anémie", they just get age of 25, before 25 they die"

Another participant felt that affected people passed away between the ages of 14-18 years. Even though it is known that a high number of deaths in SCD occur at a young age (Modell & Darlison, 2008), average life expectancy, as reported in the USA, can be up to late forties if properly treated (Platt *et al.*, 1994). In SA, while life expectancy is not as well documented, patients are expected to survive into adulthood, as evident by the existence of adult SCD services at, for example, Groote Schuur Hospital in Cape Town, which is involved in the follow up of more than 50 adults SCD patients.

Data on life expectancy in the rest of Africa is similarly restricted, with estimates suggesting childhood mortality rates between 50-80% (Weatherall, Akinyanju, Fucharoen, Olivieri, & Musgrove, 2006). Recently, Makani *et al* (2011) conducted the first study to present mortality of SCA in Africa among a group of patients in Tanzania. The study reported mortality rates similar to those found in the USA before the use of penicillin prophylaxis, 1.9 per 100 person years of observation, but revealed that this was likely to be an underestimate, as the study was conducted at a university teaching hospital where patients had good access to health care (Makani, Cox *et al.*, 2011). The highest incidence of death in that study was found during the first five years of life. Further evidence to support early mortality among SCD patients in Africa is the fact that in a randomly selected Cameroonian cohort of approximately 600 SCD patients, less than 5% were more than 40 years old (Wonkam, Ngo Bitoungui, *et al.*, 2014)

Even without exact numbers, it would seem several participants in this study had experienced similarly increased mortality rates in their respective countries, resulting in their expectation of significantly reduced life expectancy for their child.

Several participants expressed the hope of their child surviving beyond the years they thought of as normal, while others seemed more inclined to accept than hope. Examples of both these outlooks can be seen in the following quotations:

P9M: “For my son, I hope they going to pass that age. Yes that, I believe that.”

P2F: “I just accept it, because at the end of the day we all have to die. He's 10 now already. If he dies... I made peace.”

To further express the ingrained belief of reduced life expectancy, some participants conveyed the views of certain people from their country that the early demise of a child affected with SCD was so certain that treatment became almost pointless in their eyes, as is shown in the following quotation describing community member's attitudes:

P7M: “If she is already sick you don't have to spend a lot of money to treat her and all these things. You can just....you see.”

A study in Ghana reported similar findings, wherein parents were encouraged to ‘wait patiently for the day of their child's death’ (Dennis-antwi, Culley, & Hiles, 2011).

3.3.4 Knowledge on inheritance of Sickle Cell Disease

Most participants had some idea that SCD was inherited although none used the word inheritance. Rather the condition was described as ‘being in the family’ or ‘being in the blood’. This last description could be somewhat confusing, as it could also refer to the cause of SCD being literally in the blood. The indication that the phrase was being used to refer to inheritance was deduced from the context, as shown in the following quote:

P14F: “Then he (the doctor) explained to me, this sickness it comes from the blood. Example, you and your husband make the same blood, like that.”

About half of participants (n=8) correctly inferred the autosomal inheritance pattern of SCD, by stating that it is inherited from both the mother and father. In comparison, in a group of

Cameroonian patients, almost all participants defined SCA as a blood disorder (100%, n=130), inherited from both the father and mother (93%, n=120) (Wonkam *et al.*, 2011).

A few participants of this study did not know or were unsure whether SCD was inherited, while a couple of participants knew it was inherited but were unclear on the exact mode of inheritance:

P3F: "It may come from the mom only or from the dad only or from both of them."

In two cases, participants had assumed that the disease would be inherited from the parent who also had family members affected. In other words, if the father had a family history of SCD, the disorder had been inherited from him, and vice versa. In both these cases however, it was the parent not present at the interview who had had the family history, and could therefore be seen as the 'one to blame' for the child inheriting it. This is one example of how knowledge can be manipulated, consciously or unconsciously, to lessen guilt and influence denial, a phenomenon that will be discussed in more detail in a Chapter Four.

Several participants seemed to accept that as long as someone in either parent's family had previously had SCD, the child was at risk of being affected. The opposite is however also true, with a few participants believing that if no family members were previously affected, their child could not possibly be affected with SCD.

Long *et al.* (2011) described similar misconceptions, such as the belief that only girls are affected, as individuals try to make sense of the SCD pattern of inheritance seen within their family. In a study among carriers of SCD, with and without affected children, Acharya, Walsh Lang, and Friedman Ross (2009) reported a mean score of 68% on a questionnaire designed to measure knowledge of inheritance of SCD. The difficulty in knowing and understanding the inheritance of SCD appears to extend to several populations.

Explaining inheritance can be difficult, especially in cases where English is the 2nd or 3rd language, and the terms are unfamiliar. Doctors and genetic counsellors alike often try to clarify the concept of inheritance by using alternate phrases such as 'runs in the family'. However, describing an inherited disease as 'in the family' does create the misperception that if it's not previously seen in the family, it is not the inherited disease in question. This could be seen in this study, and is exemplified by participant P3F, who said:

“If it’s been inherited from someone, then in history, in my father’s family or my husband’s family in history, we should have heard someone had this disease before. On either side we should have known, at least, that there was someone. But that only started with this one”

The belief was very strong in some participants that the diagnosis in their child must be wrong, partly influenced by this misunderstanding of inheritance, as family history was felt to be a prerequisite to being affected with SCD. In rural Kenya, misperceptions regarding inheritance was found to reinforce blaming patterns within families, as well as contributing to the low initial recognition of SCD and its cause, which were in turn associated with poor surveillance practices (Marsh *et al.*, 2011).

3.3.5 Knowledge of cause of Sickle Cell Disease

Quite a number of the participants did not know the precise cause of SCD, although the majority identified it as ‘something wrong’ with the blood. Descriptions of what exactly was wrong in the blood varied greatly as is illustrated by the following quotations:

P17F: “Sickle cell is about good “sanguine”. I don’t know the English name. It’s about the blood.”

P6M: “It is a genetical disease, inherited genetical disease, that causes change of the red blood cells in my son’s body.....The abnormal function of the red blood cells, sometimes, the haemoglobin is low and it gets stuck into the body.”

Knowledge ranged from knowing only it had something to do with blood, right through to knowing it was caused by genes that lead to abnormally shaped red blood cells. Perhaps uniquely among similar African studies (Dennis-antwi *et al.*, 2011; Ebrahim *et al.*, 2010; Nzewi, 2001; Ohaeri & Shokundi, 2001; Treadwell, Anie, Grant, Ofori-acquah, & Ohene-frempong, 2014), no mention was made of a possible supernatural cause for the disease. These findings suggest an improvement in the knowledge of the condition in sub-Saharan Africa. Indeed, SCA

was known in some parts of Africa before the 20th century with disease specific names that evoke acute, painful episodes or reincarnation beliefs (Nzewi, 2001; Onwubalili, 1983).

Several participants, giving a bit more information on what they thought was wrong with the blood of a person with SCD used phrases such as ‘shortage of blood’ and ‘low blood’ to describe the cause of SCD. This could be a description of anaemia, which is more accurately a symptom of the disease, and as it causes the child to become pale can be construed as not enough blood. This assumption that SCD is caused by a shortage of blood could also be due to the treatment thereof with transfusion, as illustrated by the quotation:

P16F: “I know that it’s because they have a shortage of blood, and sometimes are waiting for transfusion.”

In addition to P6M, quoted previously as giving an almost perfect description of the cause of SCD, two more participants knew that the cause was related to the shape of blood cells. Participants P3F and P11M correctly stated that SCD was caused by abnormally shaped blood cells, although they were mistaken as to what exactly that shape was. P11M described it as follows:

“Sickle cell means they cannot manufacture blood like this, because their cells are semi-circle. They are not like your cells which are normal, which are complete.”

It was participant P3F who had a particularly interesting take on the abnormal shape of the cell:

“What I understand, they say it’s when the blood cells have an S shape and does not allow for enough blood to run through.”

This assumption on the shape of the red blood cells was probably made on account of the disorder often being called ‘SS’, or affected persons being described as having ‘SS’ haemoglobin and the participant having prior knowledge to that effect. This participant was aware not only of the abnormal shape of the blood cell, but also that the shape then leads to obstruction of blood vessels.

This was not the only incident of terms or names leading to confusion among participants. Another participant, P17F stated that she knew SCD was caused by children inheriting blood from their parents, but went on to say that both she and her husband were O-positive, and she does not understand how their child could be “AS” or “SS”.

These are two examples of instances where certain terms were misinterpreted by participants and led to misunderstandings. In the case of participant P3F, the misunderstanding was quite harmless, albeit interesting. In case of participant P17F, however, there was significant confusion that could have been avoided if the terms were explained more clearly. ‘AS’ and ‘SS’ are often used by health care professionals, as they are short, easy, and often familiar. But familiarity does not necessarily equal understanding, and it is important that even these ‘easy’ terms are clearly explained and clarified to parents to avoid adding to their confusion.

Another participant said it was possibly caused by “*something in the bones*”, an assumption most probably based on the fact that the pain occurs in the joints and bone and is a tangible symptom in a child affected with SCD.

Interestingly, two patients thought SCD might be caused by malaria mosquitoes. Both participants knew that there was a link between SCD and malaria, and that it was found more often in places where malaria is common:

P10F: “Sickle cell has, sometimes, like malaria. So maybe when, that time, the mosquito...”

Interviewer: “So it comes from mosquitoes? Like malaria?”

P10F: “Maybe, yes.”

As discussed in the literature review, the high prevalence of HbS carriers, and consequent individuals affected with SCD, arose due to an evolutionary advantage against malaria. So while the cause and effect of mosquitoes and SCD is not as direct as this participant thinks, it does exist.

3.4 Chapter summary

This chapter discussed the participants’ knowledge of SCD on different topics related to the illness. It was found that prior knowledge of SCD significantly influenced parent’s expectation

of the disease, both with regards to its physical appearance and outcome. Often the absence of what certain participants thought of as a SCD ‘look’, specifically those observed in DRC where most participants came from, gave rise to doubts about the diagnosis. Several participants voiced very exact ideas on the likely age of survival of an affected person, as based on their experiences in their native country, all considerably less than is expected in SA. While majority of participants knew that SCD was inherited in some way, knowledge about the exact pattern of inheritance varied, with participants drawing conclusions from their experiences of the disease. This sometimes led to the misconception that SCD could not be inherited without the presence of an affected family member, or that the disease was inherited only from the parent with a family history thereof. Knowledge of the exact cause of SCD was limited, with most participants knowing only that it was a problem with blood. A visual summary of this information is presented in Figure 3.1. A discussion on how this knowledge can be influenced and enhanced through genetic counselling will be presented in Chapter Five. The following chapter will discuss the findings on participants’ experiences in raising an affected child, including the burdens they face and the methods they employ in order to cope with their circumstances.

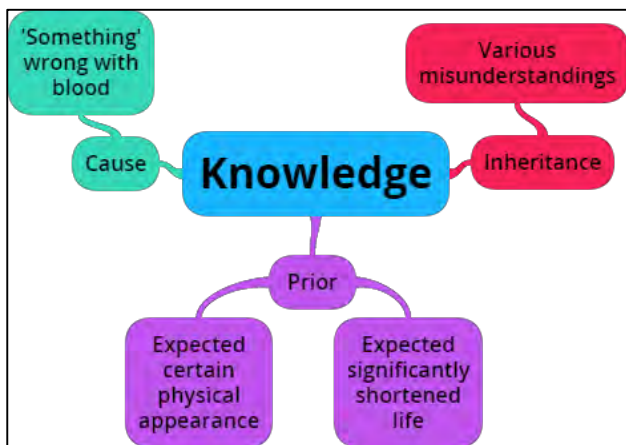


Figure 3.1: Summary of findings on knowledge of SCD

Chapter 4: Results and Discussion of Participants' Experiences

4.1 Chapter introduction

This chapter will present and discuss various aspects of participants' experiences of caring for a child affected with SCD. These experiences, as described by participants, can be perceived as burdens to the parents, and have been divided into two groups, those with practical implications for the parents and those with psychosocial implications. Coping strategies and psychological defences employed by participants to overcome these burdens will also be explored and discussed.

4.2 Practical implications

4.2.1 Ensuring good nutrition

The subject that arose most often as a complication of raising a child with SCD was that of nutrition. All participants, in one context or another, mentioned nutrition related to their affected child, discussing either the expense of high quality food, or the difficulty of making their child eat as much as is needed.

Although persons affected with SCD do not have any special dietary requirements, studies over the last decade have shown the presence of micro- and macronutrient deficiency among affected individuals, despite adequate dietary intake (Hyacinth *et al.*, 2013; Hyacinth, Gee, & Hibbert, 2010). This deficiency is thought to be due to the increased nutritional demand required by the disorder, and could explain why children affected with SCD have a tendency toward malnutrition (Hyacinth *et al.*, 2013).

As a result of this tendency toward malnutrition, some participants described difficult experiences relating to ensuring nutrition in their child. Participant P8F described a time before her child was diagnosed as follows:

“After the time, when he was a baby, he’s a small, small baby. Not eating. Most of the time he sleeping. And I take him to the doctor clinic and they shouting at me, why you don’t feed your child properly? Why you don’t treat him properly? I was so

surprised and I tried to explain to them, I don't know what's going on."

This experience, as shown by the participant's animated language, had apparently greatly impacted her and she went on to describe how she always ensures her child has 100% fruit juice and porridge, even at times when there is no money, and she herself has to do without some necessities.

Many other participants also mentioned the quality of food that they tried to procure for the affected child, highlighting the importance of what they call '*nice food*'. Among those mentioned were things like full cream yoghurt, cheese, vegetables and mealie meal (a type of flour used to make porridge). Many participants mentioned the benefit of ensuring good nutrition and hydration, stating that in their experience it improved overall health. However, while most realised the need for this, a few also stated the difficulty and expense of ensuring such high quality food at all times.

Apart from the expense, there is another factor related to good nutrition that many participants experienced as problematic, namely implementing it. Many participants indicated that their affected child is often unwilling to eat, as illustrated by the following quotations:

P11M: "The hardest part is the way it... he doesn't want to eat, and you will be forcing every day. He doesn't feel like eating anything, he just eats a little bit and says he's all right. And if you don't eat and drink enough water, for them, it becomes a problem. But he doesn't want to. You can't force him to eat. There is nothing you can do."

P14F: "Sometimes you give them something with the diet. For the tablet, you see. And I can shout, drink, this is your life. If you don't drink this you going to die. It's difficult."

Both participants quoted above, and others, came across the same dilemma, with regards to forcing a child to eat when they are unwilling. Participant P11M had come to the conclusion that nothing could be done without upsetting the child unnecessarily.

Judging by the intensity of her statement, participant P14F seems to experience this problem on a much deeper level. This could be as result of the severity of her child's condition, or her own perception thereof. It is clear that she is very aware, on a daily basis that her child could die at any moment. "Thoughts of death at any time" was reported by 55% of Cameroonian parents with SCD-affected children as occurring "frequently or all the time" (Wonkam, Zameyo Mba *et al.*, 2014). Such frequent mindfulness of the possibility of death might have a negative psychological effect, not only on a parent, but also the child.

This participant's quote illustrates one other fact about nutrition shared by a few others: ensuring the child eats before taking their medication. A number of participants mentioned that they found their child to be less likely to suffer from side effects if they take their medication after a meal. This direct correlation between eating and health serves to intensify the burden experienced by participants around ensuring and enforcing good nutrition, despite the cost thereof.

4.2.2 Financial stressors

In addition to the cost of high quality food, there were several other factors contributing to the financial burden experienced by participants. While the cost of having a child affected with SCD has been well described (Brown *et al.*, 2010; Ievers-Landis *et al.*, 2001), it has not been done in the South African health care context. Many participants described one significant factor that influences the cost of having an affected child in SA when compared to their country of origin. That is the fact that treatment in SA can be accessed through state hospitals, and the cost thereof is covered by a national health system. Through this national health system, treatment is available whenever it is needed, and patients are then billed a percentage of the cost according to their income. This is different from the arrangement in, for example Zimbabwe, as described by participant P11M:

"There is a great difference, because you see, the medication in our country, sometimes I need to pay it there. If I don't have the money he won't get treated. But here he can be treated and I can pay you later on, when I have got the money. So unlike here, if my medication is finished, I come and take. And I can come and pay half of that money, or just a little bit, and I

carry on. So as much as I can afford for that time. But they don't stop me from coming here with the boy."

This quote shows not only the difference in health care systems, but is also a practical example of the economic support described by many of the participants as a crucial advantage of South African health care. Several participants from DRC also described SCD as being 'very expensive' in their native country, while being significantly less so in SA. Nevertheless, even though cost is subsidised and can be paid over a period of time, it is still an extra expense to be planned and provided for.

Furthermore, although treatment is fairly inexpensive, there are many other aspects of having an affected child that can be very costly. For example, patients need to be brought to hospital frequently and the commute can be expensive, as it often involves the use of public transports such as taxi's or buses. Depending on where participants live in relation to the hospital and how often the affected child needs to come to hospital, this can become a considerable expense. This is illustrated by the following quotation:

P17F: "I don't have money, you see. Like, there are nights, I don't have money, so I must soothe him until morning. It was so difficult for me, because any time he get sick, so if you don't have money and if it's really late..."

This describes not only the perception of travel cost as great, but also illustrates how cost of travel can have a negative impact on the health of the child, resulting in him or her being hospitalized later than is prudent. After a number of experiences such as described above, participant P17F ultimately decided to relocate to within walking distance of RCWMCH, where she now has easy access even at night.

A less direct financial stressor related to an affected child is the resultant loss of income due to missing work. Reduced carer employability in the case of a SCD affected child has been described before (Ievers-Landis *et al.*, 2001), and seems to be supported in this study by a statement made by participant P2F:

"At the beginning I was working, but now when he got so sick I had to stop working. And now, I'm not working since ten years."

Participant P2F was the only participant in this study that directly related her son's illness to her being unemployed. However, she is also one of only two participants in this study who is unmarried, and therefore solely responsible for the care of said child.

Previous studies, discussed in the literature review, revealed many instances of loss of income due to missing work (Brown *et al.*, 2010; Moskowitz *et al.*, 2007; Wonkam, Zameyo Mba *et al.*, 2014). However, as many participants were unemployed, it did not emerge as a strong theme in this study. Still, it cannot be denied that many participants felt a significant financial burden, possibly intensified by the high level of unemployment. It is perhaps best summarised by the following quotation by participant P2F:

"I'm still coping. But it does make...there's a hole, like a big hole in the pocket."

4.2.3 Caregiving time

As discussed in the literature review, caring for a child with SCD can be time consuming (Moskowitz *et al.*, 2007). This was experienced and described by participants in this study in one of two contexts: those who were employed and found working and having a chronically sick child challenging; and those who found everyday parenting tasks more time consuming than with healthy children.

In the few cases where both the participant and his/her spouse were employed full time, the child would be sent to school, when possible, or left in someone's care. Both these solutions were at times found to be less than ideal by the participants. Schools would often phone parents to collect their child at the first sign of illness, disrupting their work day, while the second possibility is hampered by the fact that the participants in question had no family in Cape Town to whom they could entrust the child's care. Participant P17F depended on her eldest child to help look after her affected sibling, an option not available to all, and very reliant on the vigilance and capability of the sibling.

Then there are the daily tasks that become more demanding when one's child is affected with SCD. A previous section already discussed the difficulty of making an unwilling child eat, but this problem can also have time implications. Participant P10F described having to cook 'special' meals for her child, both because she is more likely to eat the higher quality food and because it is too expensive to make it for the entire family. In the same line, participant P4M

described the struggle involved in getting his affected children dressed for school, with him wanting to ensure they are sufficiently warm and protected against infection, and the children becoming upset at being forced to dress differently from other children.

Even in the case of a parent being available at all times, participants found that the caregiving time involved made it difficult to give equal attention to household tasks, the affected child, and other healthy children. This last is illustrated by the following quotation:

P4M: You see, (daughter) can get sick, but I can't be worried much. But he, when he get sick, I am worried too much. It's like I don't even...It's like I just try to... you see..."

As seen in the above quotation, participant P4M found it very difficult to express himself on the subject of his healthy daughter receiving less of his attention during an illness than one of his affected children. These feelings can contribute to the considerable emotional burden of having a SCD affected child, which will be explored in the next section.

4.3 Psychosocial implications

4.3.1 Emotional burden

The emotional burden experienced by parents of children affected with SCD can be caused by a number of factors associated with the disease, some of which were discussed in the literature review. In the case of this study, uncertainty was a great contributor to parental emotional distress, both in terms of prognosis and the unpredictable nature of the illness.

As an illustration of the first, several participants mentioned their fear of an uncertain future with their child. Many revealed a constant awareness of the possibility of their child dying, and at the same time conveyed an inability to come to terms with the possibility thereof. Consider the following quotation of participant P14F:

"I'm very stressed, really, because I don't know what is going on. I think who is going to help me with my child? I'm worried, one day, this blood, maybe one day it is going to finish. And this child, is she going to grow old now? I don't know."

As shown by this quotation, the uncertainty can be influenced by several things. In the case of participant P14F, confusion about treatment played a part, as she was concerned about blood being for transfusions ‘*running out*’. She expressed further anxiety at being unsure of her present situation and her daughter’s unclear prognosis. While it is impossible to accurately predict the future of a child with SCD, this is also true of a healthy child. This mother, and those like her, might benefit from some general information about the prognosis of SCD. Even if the information contains bad news, such as the expected reduced life span, it can still be valuable by removing some of the ambiguity felt by the participant. Baldwin and Carlisle, as cited by Atkin *et al.* (1998) found appropriate information was able to facilitate coping in parents, as it helped them make sense of the condition (Atkin, Ahmad, & Anionwu, 1998).

Something else that caused considerable emotional distress among participants was the expectation and occurrence of pain crises. As pain crises occur randomly they cannot be prepared for, and this was described as being very unsettling and the cause of much instability in everyday life. In addition to this disruptive quality, pain crises were described by many participants as one of the most challenging aspects of the disorder to witness and handle on an emotional level. This was also found in a Canadian study on mothers of SCD affected children, where all participants reported constant fear of their child’s sudden hospitalisation or possible death. The study further reported participants feeling a lack of control over their everyday life due to the unpredictability of pain crises, leaving them with a persistent feeling of helplessness (Burnes *et al.*, 2008). Participant P4M, recalling his feelings during a crisis, gives a sense of this helplessness and hopelessness in the following quotation:

“We just don’t feel to...to do... anything. We don’t feel like...like we just feeling nothing is better in this world. I can try to explain, you just feel like we have something...we carrying something. A heavy staff that we don’t know how to pick up. So we just carry that heavy staff and then we don’t know who can help. We just feel this staff is very heavy to us, but still we must have hope, maybe we going to get help.”

Participant P11M said that he often wished he could be the one who was suffering from pain in his child’s place. Other participants had similar feelings, citing their inability to relieve their child’s pain as ‘the hardest part’ of having an affected child. In a recent Cameroonian study, pain crises were found to be the SCD disease related stressor that most undermined the coping ability

of caregivers, with the unpredictable nature of the crises possibly contributing to the perceived burden thereof (Wonkam, Zameyo Mba *et al.*, 2014). Participant P11M, quoted above, went on to describe the ‘suffering’ as now being part of their everyday life, and something that both he and his child will have to come to terms with eventually.

Contributing to feelings of hopelessness was the incurable nature of SCD, causing a few participants to liken it to other ‘incurable’ diseases:

P17F: “There’s no healing. Like for malaria, the malaria is finished. But not for this one, understand? It’s like cancer, it’s like HIV. It’s the same because it’s not healing... I have no peace in my heart. You know they are to leave you.”

All the factors discussed in this section can add considerably to feelings of anxiety, distress and depression. In order to overcome or manage these feelings, many participants adopted various coping styles and psychological defences, which will be discussed in section 4.4.

4.3.2 Stigma

Stigma is defined by the Oxford Dictionary as a mark of disgrace associated with a particular circumstance, quality, or person. In the case of this study, the circumstance would be having a child who is affected with SCD. Stigma with regards to SCD has been reported before, and often stems from uneducated ideas of SCD, such that it is contagious or a curse (Burnes *et al.*, 2008; Anie & Green, 2012). In this group of participants, the majority had not experienced any stigma related to having an affected child, although those who had, had found it to be a deeply distressing event.

As SCD is a fairly unknown disease in most South African communities, it is not surprising that most participants had never felt judged for having a child affected with it. In fact, several of the participants who had experienced stigma found it to be unrelated to SCD specifically, but rather a result of community members noticing that the child was constantly sick. This led them to the conclusion that the child, and therefore likely the parents, were HIV positive.

An example of this can be found in the following quotation:

P4M: "People have to talk. Some people they think maybe HIV. You try and explain them, they will not really understand it. They will think you are bringing other stories. So it is not easy to be in this kind of business."

Being HIV positive in SA still carries significant stigmatisation (Skinner & Mfecane, 2004), and it would appear that judgement also extends to other 'unfamiliar' illnesses. A few participants, including P4M, had found that the frequent clinic appointments and hospitalisation of their child was presumed by community members to be a result of them being HIV positive. As also illustrated by the quote, most found it unhelpful to deny or correct these assumptions, as it was commonly interpreted as evasiveness due to shame about their HIV status.

In an isolated case of stigma experienced by one participant, the object of judgement was once again not SCD itself, but rather the treatment thereof. Participant P17F had been told that blood transfusions were unnatural, and that her child was 'a dead person, not a child'. Both these instances of stigmatisation could be resolved or reduced through community education to create awareness of SCD.

Those participants who had experienced censure related more specifically to SCD seemed to have encountered it mostly among individuals who had some experience of SCD, for example family members or individuals met in their country of origin. These individuals often had knowledge and exposure to SCD, and therefore were prejudiced against it. Some examples of judgement are described in the following quotation:

P7M: "You see, many people, if you got a child with SS, they are going to laugh at you, you understand. Because many people they put in their minds that the child is going to die. When they see your child they got SS, they say 'ah this child, she is not going to last. She is going to pass away. She is going to die'. They always think about these things. And already they putting something in your mind. Like you knew this person, she is going to pass away one day, you don't have to call us and worry about, if she sick. If she is already sick you don't have to spend a lot of money to treat her and all these things. You can just....you see."

Similar experiences were described by other participants, also related to the expectation of the early death of his affected child. The infrequency of these events make them no less distressing, and one participant ultimately estranged herself from those members of her family who had ‘given her pain’ because of her child having SCD.

4.3.3 Support

A good support system can often help reduce the emotional, and even financial, burden experienced by caregivers of children with SCD (Brown *et al.*, 2010). Unfortunately, when participants spoke of social support, it was mostly to comment on their lack thereof. In a few cases, this was a direct result of having an affected child.

In the case of participant P15F, the relationship she had had with her child’s father was ended after they realised the risk of having more affected children. She is now a single mother who had the following to say when asked about how she copes on her own:

“I never discuss about... Even at school my friends don’t know, I don’t discuss about that. I’m not feeling free to talk about that. I don’t want to put it in my head, I get sick. Otherwise, I’m also going to fall down.”

She went on to say that emotional strength is essential if you have a child with SCD. Her dependence on her own strength was partly brought about by the fact that she has no family in SA, and no one she could feel ‘free’ with.

As almost all participants were originally not from SA, absence of familial support was a common occurrence. Moreover, a few participants felt that they would be unable to return to their country, and family, as they would be unable to afford the same level of care for their affected child. As previously discussed, SCD treatment at state hospitals in SA is inexpensive, of a good quality and readily available (Wonkam *et al.*, 2012). This benefit was considered worth staying for, even by those who had not intended to. It was well put by participant P10F, who when asked if she ever thought about returning to Malawi replied:

“I can’t. I need, but I can’t. Always very difficult being here.”

Family is, of course, not the only source of social support. A few participants mentioned friends as ‘helping out’, but would go on to say that they did not feel comfortable telling friends of their

child illness. Often participants had no support in managing their child's illness other than that of their spouse. Fortunately, in the majority of cases, participants described their spouses as being extremely supportive. This was very well expressed by participant P4M:

“We need support, because we decided we have to be one. We have to be one, with these kids. I’m her counsellor, she’s my counsellor.”

However, to increase the support available to participants, other support structures could be put in place. There is currently no support group active in Cape Town, or SA, for parents or individuals affected by SCD, but they have proven to be very successful in alleviating emotional and psychosocial stress and now play an integral part in the SCD care programs of the USA and UK (Dennis-antwi *et al.*, 2011; Dennis-antwi, Dyson, & Ohene-Frempong, 2008). The forming of such a group in Cape Town could benefit many participants in this study and others like them by giving them much needed support as well as the opportunity to share their experience with others going through a similar situation.

4.4 Coping with Sickle Cell Disease

Given the emotional demands of raising a child affected with SCD, parents employ a variety of methods to help them cope with their child's diagnosis and disease. Understanding these methods, utilising its strengths and minimising any threats to it, can help improve the support services provided to parents in similar circumstances (Atkin & Ahmad, 2000). This section will explore and discuss the coping strategies and psychological defences employed by participants in this study.

4.4.1 Coping strategies

Coping strategies are employed for problem solving or to change the meaning of what is being experienced (Uhlman, Schuette, & Yashar, 2009). Often, when the stressful situation is unchangeable, as in the case of a chronic illness like SCD, coping strategies tend to be more emotion focused, rather than problem solving (Weil, 2000). This was found to be true for most participants.

As an example of an emotion focused coping strategy, many participants used religion to help them manage their circumstances, describing it as giving them the strength and ability to cope

with their child's diagnosis and illness as well as providing a source of hope. Atkin and Ahmad (2000), in a study on parental coping with haemoglobinopathies, found that religion was a key resource in helping parents cope with the diagnosis and symptoms of SCD. In this group of participants, where many had no family to offer support, many turned to God as their main source of strength and emotional support.

Some benefits of religion as a support structure are illustrated in the following quotations:

P14F: "You can't help me every day. Only God can help me. Jehovah he give me strength."

P4M: "We always put our hand in God, and put our children in God, because the doctor just only do what he learned in school, but God he will give everything after doctor finish."

P8F: "I have also pray to God, God must give the doctor more and He must also help my child. You know what I like to say, God mustn't disappoint me for my child."

The first quotation highlights the very important fact that God is the only support available at all times, a very valuable quality when it comes to a disorder such as SCD, which can present with problems at any time.

The last two quotes, while showing the trust the respective participants had in God also demonstrated something else: the importance of religion in conjunction with other coping strategies. In this case the more problem solving focused strategy of embracing the medical model. Embracing the medical model is defined as the acceptance of the medical definition of the disorder and using this information as a means to acquire some control over the situation (Atkin & Ahmad, 2000). Both participants, P4M and P8F, quoted above relied on both religion and medical expertise to take care of their child, using one to strengthen the other.

However, this conjunction of strategies was not found in all participants, with many using religion as a means of escape-avoidance. Escape-avoidance as a coping strategy was identified by Lazarus in 1991 (Uhlman *et al.*, 2009) and is summarized by Uhlman *et al* (2009) as hoping for a miracle. For many participants, relying on religion had surpassed merely drawing strength and support from God, as is evident from the following quotations:

P17F: "I know God will heal him, God is able to do something for him, I know that. God will do something. I believe in Jesus' name. It is so painful....I know God is going to change his blood."

P5F: "God will change everything. Because this disease, I don't like it. I said, God, there is no sickness or disease He cannot heal. That's why I asked God to please intervene for me in this. I believe in God to intervene. Nothing is impossible for Him, because I can't accept it."

Some participants, those quoted above included, clearly hoped for their child to be miraculously healed. While this hope is understandable and normal under the circumstances, some participants seemed to expect it with a worrying naiveté. The complete disregard of any other source of hope could lead to continued disappointment as the child fails to be healed, and in the long run could even cause the participants to question their faith.

4.4.2 Psychological defences

It's difficult to separate coping strategies from psychological defences, as they are closely related. There is however one significant difference. While coping strategies involves conscious activities that aim to reduce emotional distress, psychological defences are an unconscious mental process by which painful or unacceptable realities are kept from reaching consciousness (Weil, 2000). Psychological defences are essential for maintaining healthy mental functioning.

While there is no universally agreed upon list of psychological defences, some are commonly accepted and well defined (Weil, 2000). Denial is one of the most universally experienced forms of psychological defences, and was also found in a substantial number of participants in this study. As described by Uhlman *et al* (2009), denial is the inability to acknowledge to oneself certain information. It is a common response when the information, in this case a diagnosis of SCD, elicits shock and fear. When it occurs at an appropriate intensity, denial can keep individuals from being vulnerable to serious psychological harm, but, because denial in essence distorts perception of reality, it can contribute to psychological dysfunction when overly strong and rigid (Weil, 2000). The following quotations illustrate two examples of denial:

P9M: “And let me tell you the truth, at first, when they notice that (referring to son having SCA), I didn’t believe them. I didn’t believe that.”

P3F: “I don’t really believe in it. And I’m not angry for that and neither sad for that. I just don’t believe in it.”

The first participant quoted experienced denial upon first finding out about his son’s diagnosis. The shock of an unexpected diagnosis as well as the imminent loss of the ‘healthy child’ resulted in a temporary disbelief of the information provided. But denial is usually short term (Uhlman *et al.*, 2009) and in the case of participant P9M, as well as a few others, he came to accept the diagnosis after a time, assisted by a genetic counselling session.

However, as can be seen in the second quotation, this was not the case for all participants experiencing denial, and in a few, the denial was persistent and seemed to have reached a level bordering on dysfunction. Even after several years, participant P3F (previously quoted above) maintained her disbelief in the diagnosis:

“All I think is one day it will be proven wrong. Because I don’t believe it is inherited, I don’t believe it. I will never believe it at all. Not, like, not believe in this sickness, but believe that my kids are sickle cells.”

This continuous denial can have a harmful impact, not only on the person experiencing it, but also others in the family. In the case of participant P3F, it had already led to some disagreement between her and her husband, as he believed they should be focused on finding a solution or cure for their children’s illness. Her failure to acknowledge some aspect of the reality apparent to her husband has seemingly distanced her from the only support she had. On the other hand, the denial had resulted in no apparent harm to her two affected children. At the time of the interview, both were being treated only with folic acid and prophylactic penicillin. This treatment was normalised by the participant through saying that anyone can take antibiotics and vitamins, in the same way that all children can develop fevers. She did not perceive either the treatment or symptoms as being proof of a SCA diagnosis.

It is important to note that in this group of participants, denial was often reinforced by their prior knowledge about SCD. While it is natural to make assumptions based on available information,

as discussed in Chapter Three, the information, as inferred from experiences in their native country, is not always accurate or relevant to a South African context. Consider the following statements:

P17F: "I heard, in the Congo, that people have Sickle Cell. The way they look, not like my baby...I told them they are lying. He didn't look sick. No, they are lying, they didn't check nicely."

P9M: "I didn't believe, because if I saw my son and compare with all the kids from my country, I can see, like, the hair looks like my hair, not like the one from my country. But then I did get counsel here and I could understand everything."

P3F: "If it's been inherited from someone, then in history, in my father's family or my husband's family in history, we should have heard someone had this disease before... And in my country, when we look at kids who have Sickle Cell Disease, they don't look as healthy as these ones. They look sick, they are forever sick."

These participants, and others, expressed an unwillingness to believe the diagnosis because their child did not look as they expected a child with SCD to look. There are a few explanations for this difference in appearance. In the case of some participants, a diagnosis was made at a very early age, before the child had displayed any symptoms for a long period of time. But, in most cases, children were receiving optimal treatment, something not achieved in all African countries. As a result of this treatment, the children appeared, to the non-specialist eye, to be reasonably healthy and unaffected by SCD.

In addition to this difference in appearance, participant P3F's quotation illustrated another point where prior knowledge can be misunderstood and used as an argument against the diagnosis. This was the misunderstanding of the pattern of inheritance of SCD. While participant P3F knew that SCD was an inherited disorder, she assumed that it could only manifest if someone in the family had previously been affected. This belief was shared by a few other participants and led not only to a disbelief of the diagnosis, but also distrust in the doctors or health care workers

who were insisting the diagnosis was correct, without explaining how this was possible without an affected family member.

Prior knowledge can be so ingrained that in few participants it seemed to lead to what could be described as a confronting coping strategy. This is summarised as a strategy whereby a person tries to change the opinion of the people in charge (Uhlman *et al.*, 2009). At the time of the interviews, participant P3F was in the process of pursuing private SCD carrier genetic testing for herself and her husband. Despite the cost, she felt if she could bring the results to RCWMCH it would prove her belief that the doctors were wrong in their diagnosis. This challenging and almost antagonistic approach would need to be handled in a sensitive and effective manner to avoid any detrimental effects, to both the parent and the child.

4.5 Chapter summary

This chapter presented the findings on the burdens experienced by participants of this study in regards to raising and managing a child with SCD. The expense and/or effort of ensuring good nutrition was cited by all participants, while the financial strain and that of caregiving time varied according to personal circumstances. The emotional burden experienced was intensified by the uncertainty of the prognosis, as well as feelings of helplessness, especially during pain crises. Further contributing to their burden of raising an affected child was the fact that the majority of participants had limited social and emotional support. Possibly as a result of this, religion was found to be an important mechanism for coping in this group of participants. Other coping mechanisms were also discussed as well as psychological defences, in particular examples of denial. Figure 4.1 provides a visual summary of these findings. The following chapter will explore the role that genetic counselling did and could play for participants, as well as their attitudes towards screening services.

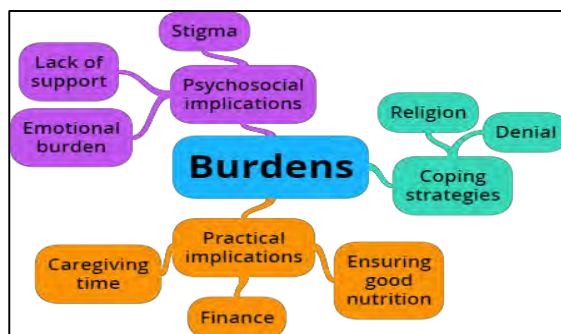


Figure 4.1: Summary of findings on burdens experienced by participants

Chapter 5: Results and Discussion on Screening and Genetic Counselling

5.1 Chapter introduction

This chapter will describe participants' opinions of the screening available for SCD, including their attitudes towards the termination of an affected pregnancy. Finally, this chapter will look at the participants' experiences of genetic counselling, the value, if any, they gained from it, and the opportunities for improving this service at RCWMCH Haematology clinic.

5.2 Sickle Cell Disease screening

In order to reduce the prevalence of SCD, many countries have started using various screening policies that could lead to decreased numbers of affected individuals. The screening is aimed at identifying at-risk couples, affected newborns, and affected pregnancies. While all these services are available in Cape Town, data on the practice and application thereof are scarce. A study done in 2012 found that while PND had been available in Cape Town since 2010, the uptake thereof was poor (Wonkam *et al*, unpublished data). Before implementing any policy it is important to know how the individuals it concerns would respond, and to that end participants were asked their opinion on premarital, neonatal, and prenatal screening.

5.2.1 Premarital carrier screening

Most participants (n=12) agreed with the idea of premarital carrier screening, on the basis that it could give one the possibility of a better future:

P16F: "To have a better future. Instead of having children who may be sick, it is better to have a better future by preventing it in all the ways we possibly can."

This quotation is a demonstration of the feelings of almost all the participants who agreed with premarital screening. One participant felt it would be dependent on a specific couple, and what they would do with the information gained from a screening test. Those few who disagreed with the principle of premarital screening did so because they thought it would be wrong to let something like SCD carrier status influence a relationship. This was neatly described by participant P2F who said:

“You can’t help who you fall in love with.”

One participant offered the interesting opinion that premarital testing would not have benefited her as she would not have believed the results. She went on to say that she thought many other people would feel the same way in the absence of an affected family member. A study on the beliefs of African-American women on SCD carrier screening found that while participants believed SCD to be a severe disease, and saw the benefits of screening, they believed themselves to be at sufficiently low risk to not need such a test (Gustafson *et al.*, 2007). While not entirely similar to the participant described above, it does indicate a shared belief in their own improbability to be a carrier of SCD.

5.2.2 Neonatal screening

Most participants (n=12) approved of the idea of neonatal screening, while indicating that parental permission would need to be obtained before testing the child. In many newborn screening programs, this is not done, as babies are tested for SCD as part of a routine newborn genetic screen. It was generally believed that early knowledge of diagnosis could possibly benefit the child in question, or at least give parents warning of the disease before the child would become seriously ill:

P12F: “I think it is a good idea, because you will know from day one. It will be an advantage to know before it’s too late, like we did.”

The value of knowledge, opportunity to prepare appropriate care, and possibility of early treatment have previously been reported as perceived benefits of neonatal screening for genetic conditions, including SCD (Long *et al.*, 2011).

The few that disagreed with this form of screening felt that it was ‘too late’ if the child was already born for testing to be beneficial or that testing without any physical indications would be a sign of ‘negative thinking’.

5.2.3 Prenatal diagnosis

The majority of the participants in this study (n=14) supported the idea of PND. In fact, two participants had undergone PND themselves, one in Cape Town due to a previous child being affected with SCD, and one in the UK, apparently as a result of ethnic-based screening in an

obstetric setting. However, both participants seemed to be somewhat unclear on what they had consented to when accepting the test. Participant P1F, who had the prenatal test in Cape Town, could not recall why an amniocentesis was done, and said only that she was told to do it by doctors. As this test is always offered as the choice of the woman in question and informed and signed consent is compulsive before the invasive obstetric procedure, this is unlikely to be the case. This participant's confusion about the test she had done highlights the importance of comprehensive pre-test counselling. The participant did however remember that the test showed her child would not be sick, which is perhaps the most important part.

Participant P2F, who was tested in the UK, had a similar experience, although she emphasised the value of a prenatal test.

"I agreed to everything. Yes, yes, yes. To be honest I was nervous and I just say yes. So I did what they were asking me and... There's nothing wrong with that you know. Because if they didn't do the test, I would have had no idea he had sickle cell. What they did was good because now I know what to prepare myself for."

Although this participant said that she agreed to be tested mostly because of the anxiety at her 'first time in a different country', she ultimately derived value from the test as it allowed her to prepare herself, both mentally and practically, for a child affected with SCD.

This opportunity to prepare oneself was in fact the reason for most participants' support of PND who felt it would be valuable to be forewarned if a child would be affected. The value of awareness was previously reported on by Long *et al.* (2011), who found that the information provided by a prenatal test was believed to ultimately benefit the pregnancy, the mother, or the family.

Of the participants who did not support PND, one cited the risk involved in the test. In all methods of invasive prenatal testing there is a small but significant risk of miscarriage (0.5-2%) (Harper, 2010). This risk was perceived as too great by participant P4M when the test was offered to him and his wife during a previous pregnancy. He described it as 'dangerous' and they declined testing.

Another participant disagreed with prenatal testing as she felt the outcome would not be actionable:

P3F: "I was asked that question before I was even pregnant with this one. I asked them what's the reason behind that. Is it that you can stop it (SCD)? They say no, you can never stop it, it is just to ask you if you would be concerned in terminating the pregnancy. So I told them I would never do such a thing."

Most participants shared this attitude toward the termination of a pregnancy affected with SCD, and would seemingly do a PND simply for the information it provided, the opportunity to prepare, and the resulting peace of mind. Few (n=4) would consider the possibility of a medical abortion.

In almost all cases this attitude towards termination was founded on religious principles. Even participant P2F, the only non-religious participant in the group, disagreed with termination for moral reasons, as shown by her response, when offered termination following a positive PND:

P2F: "Why stop the pregnancy? I can, you know, I felt him. He was here, he's still alive. He's still got all his hands and everything. And there's a reason why I got a sickle cell child. Why I want to stop it? It's a living body there."

On the other hand, there were a few participants that thought medical termination of pregnancy would be justified, considering the severity of SCD. This is shown in the following answers given by participants to the question of what they thought of medical abortion:

P17F: "This is better. It's something you didn't see, it's better than something you see, so painful. You're pregnant with this inside, if this pregnancy is not nice you can know, you can take it out. It is better than to leave someone and at nine months it is out and the baby's sick. When you see the baby, it's not so easy."

P16F: "Which is good. Which is good, actually. It's not a good condition to have a child having that for the rest of their

*life. But it is better to prevent it. Stopping it would be good.
That's according to me though."*

The first quote is the participant's explanation of how much harder it is to raise a child affected with SCD versus terminating the pregnancy before 'seeing' that child. The second quote, by participant P16F, presented much the same view, although her last sentence highlighted a very important point. She conceded that not everyone would share her opinion and in doing so allowed for a universal truth around medical termination. In the case of such a highly personal decision, influenced by many factors (from religion to personal experience), it is essential to realise that there is no correct decision. The role of the genetic counsellor is simply to offer the choice and then provide support where possible and appropriate.

Intriguingly, participant P16F had based her feeling on termination of pregnancy due to severity, and all prevention of SCD, not on her experience with her affected child, but on her previous knowledge of the disease. At the time of her interview, her child had been recently diagnosed and generally in good health. As a result she had experienced only minimal burdens of raising a child affected with SCD. However, she had knowledge of SCD from DRC, and this possibly led to her strong feelings in favour of prevention. Her attitude may also have been influenced by lingering shock at the diagnosis and fear of the future. Participant P6M was ambivalent, saying that at this time he did not know how he felt about the termination of an affected pregnancy. He could find merit in arguments made both for and against it and was unsure what he would do if faced with such a situation.

Recent studies done in Cameroon reported 40.9 and 62.5% acceptance of the principle of termination of a SCD affected pregnancy among patients and parents, respectively (Wonkam, de Vries, *et al.*, 2014; Wonkam *et al.*, 2011). This is considerably higher than the 23.5% found in this study, despite the fact that religious affiliation, the main reason for disagreeing with termination in this study, was equally high in all study groups. The difference in opinions could possibly be accounted for by another variable, difference in disease burden. With carrier frequencies ranging from 8-34% and no medical health insurance coverage (Wonkam *et al.*, 2011), SCD burden is considerably worse than in SA.

5.3 Genetic counselling

Genetic counselling, as defined by Genetic Counsellors of SA, is a medical service that provides information about genetics, helps individuals understand the impact of a genetic diagnosis on

their life, and assists individuals with making informed personal and medical decisions (www.geneticcounselling.co.za).

Parents from the RCWMCH Haematology service can be referred for genetic counselling at the discretion of the doctor at any time, although it is most often done following a new diagnosis. In recent years, when possible and the number of genetic counsellors allow it, a counsellor has been available during Haematology clinic hours for any new patients, those who had never had genetic counselling, or those the doctors felt would benefit from it. Nevertheless, not all patients are seen by a genetic counsellor and those who are, are typically seen only once unless another session is requested by the counsellor, parent, or doctor. During genetic counselling sessions the counsellors usually discuss the cause of SCD, autosomal recessive inheritance, and the implications thereof and risk to other and/or future children. Further topics may be covered on a needs basis as indicated by the parent during the session. Sessions are on average 45 minutes long.

Records of genetic counselling sessions are kept in patient folders at RCWMCH Haematology clinic, and these folders were examined in order to determine which participants had received counselling, when that counselling had occurred, and who had attended the session.

5.3.1 Influence of genetic counselling on knowledge of Sickle Cell Disease

Several of the participants (n=8) had had genetic counselling before being interviewed for this study. All sessions took place between 2007 and 2011, with the exception of participant P7M.

Participant P7M received counselling immediately before being interviewed. No amount of time had passed, so it was impossible to ascertain how well he had retained the information discussed during the session. Furthermore, he did not directly answer any questions asked about cause or inheritance of SCD. Instead, he referred to a sheet of paper given to him by the counsellor. For these reasons, P7M is not included in the following discussion.

All participants were asked if they had ever received genetic counselling, to which only three (P9M, P12F and P17F) responded in the positive. Remaining participants replied negatively or said that they could not remember. Mostly, those who had had counselling, could not recall it during the interview. This can be due to the time passing because in even the most recent cases it had been approximately four years since the genetic counselling session had taken place.

Another explanation for this lack of recall could be the fact that genetic counselling is not yet well known to parents and participants might not have remembered it as something distinct from a routine doctors' visit.

In discussing the participants' knowledge on SCD, including its cause and inheritance pattern, it is important to take into account which participants had had genetic counselling prior to being interviewed, and what the influence of that counselling was on the level of knowledge.

If the success of the genetic counselling session, for the purpose of this discussion, is seen as ability to recall information provided, success varied greatly between participants. As illustrated by the following quote, participant P6M benefited greatly from the genetic counselling session in terms of understanding the disease, despite the fact that he could not recall having a session:

“It is a genetical disease, inherited genetical disease, that causes change of the red blood cells in my son's body.....The abnormal function of the red blood cells, sometimes, the haemoglobin is low and it gets stuck into the body.”

This knowledge was in turn used to inform the child's preschool and certain family members of exactly what was going on. The preschool was concerned that the child might be contagious, but was reassured by the information given to them by P6M. This is a good practical example of how knowledge can benefit the affected child.

Another example of the benefit of genetic counselling was illustrated by participant P9M. He explained that upon first hearing the diagnosis, he did not believe it to be true. This belief was the result of his affected child not looking the way he believed a child affected with SCD should look. However, after the genetic counselling he said he 'could understand everything' and had accepted the diagnosis. Genetic counselling had helped him overcome some emotional difficulties related to dealing with his child's diagnosis.

5.3.2 Further counselling needs

Even among participants who had previously received genetic counselling, there were indications of a need for more counselling. Consider the following quotation:

P17F: “Or maybe I, with this one, I suspect, maybe I take... I move too much when pregnant. So it (SCD) comes. It’s not nice for me. I mean suddenly I think...”

Despite having been counselled on the cause of SCD, participant P17F still had some reservations about possibly causing her child’s illness. Referrals to the genetic counsellor are most commonly made following a new diagnosis, at which time parents are often overwhelmed by not only new information, but also the emotions associated with it (Atkin *et al.*, 1998). And while genetic counselling is undeniably important in order to give and explain relevant information at that crucial time, it is hardly surprising that parents do not recall all the details. Follow up sessions could help with retaining information, while also giving parents an opportunity to ask questions after having had some time to think about it.

Of the participants that had not received counselling, two were able to provide reasons. P4M explained that he did not often bring his children to clinic, and as such had not really had the chance to accompany his wife to a genetic counselling session. This is something he seems to regret and almost resent, as illustrated by the following quote:

P4M: “I’m sorry I don’t understand the point for this sickness, because I need like someone to explain it or to show me what is that cause. They just only give me the few sentence, they don’t even go through and show me what is the real cause of that sickness. I never even get a chance to speak to the counsellor, only my wife who always get a chance.”

When asked what his wife had told him about the information discussed during her genetic counselling session, he responded that she could not really explain it. It is very rare that a couple comes to the clinic together, as the clinic takes place during work hours. Most commonly, one or of the parents, or even a third party, will accompany the child to the clinic. Under these circumstances, it is understandable that often only one parent will be seen by a genetic counsellor.

However, practices can be put in place to ensure the spouse or partner receives as much information as possible. For example, the genetic counsellors have developed a SCD information sheet that briefly covers topics like inheritance and cause. This is given to parents on a need basis, but should perhaps become routine practice. Then, even if the parent that is seen

understands everything discussed, the information sheet can serve as a memory aid when explaining what was discussed during the session to their partner. Or in cases where the absent parent might be the one with clearer understanding, the information sheet can give them the knowledge they need and might have been unable to acquire otherwise.

Participant P14F had an almost contrasting reason for not receiving counselling, she did not want to. She felt that seeing a genetic counsellor would be too much for her to handle. She had never seen a genetic counsellor, and as she was very emotional in the interview, she was asked if she would be like to be referred to a counsellor. Her answer is given here:

“Sometimes I don’t want to talk. When someone asks me I don’t want to. It gives me stress.”

This was very illuminating, as it points to a misunderstanding of what a genetic counsellor is. While it is true that genetic counsellors offer emotional support, their main role would be providing information and identifying possible areas of concern. Participant P14F did not seem adverse to receiving information, but she was opposed to being forced to talk about her child being ill. She felt it would add to her stress and not improve anything. In fact, she did not want to see a counsellor, and failed to realise that a counsellor and genetic counsellor offer different services.

This participant, and others who expressed similar feelings, could benefit from a number of genetic counselling sessions, instead of only one as is routine. This will give them time to process and come to terms with all the information. It will also be beneficial if a relationship of trust is built between them and the counsellor in order to optimise the way they receive the information. It will allow the counsellor the freedom to not give all the information in one session. In this way the first session could, for example, focus only on explaining autosomal recessive inheritance, and how it might have happened that SCD has never been found in the family before. Another session can focus on explaining the diagnosis, tests, and prognosis, if necessary, and also discussing treatment.

All these things could help parents come to terms with the diagnosis, which will in turn have a positive impact on the entire family. This will also place the genetic counsellor in a position to assess if and when psychological intervention might be justified, and then make the necessary referrals.

Several participants who had not previously received genetic counselling were referred after their interview as they expressed a wish for more information on SCD.

5.4 Chapter summary

This chapter discussed participants' views on screening for SCD as well as their experience with genetic counselling. Overall, the majority of participants supported all methods of screening for SCD, even in instances where participants would not make use of specific services, they agreed that it should be available to those who chose to utilise it. Despite the severity of the condition, most participants stated that they would not consider terminating an affected pregnancy, based on religious principles. These findings are visually presented in Figure 5.1. While most participants had never received genetic counselling, some of those who did revealed increased knowledge and acceptance of SCD. However, there is indication for improvement of this service, the most valuable of which could be offering follow-up sessions to all parents.

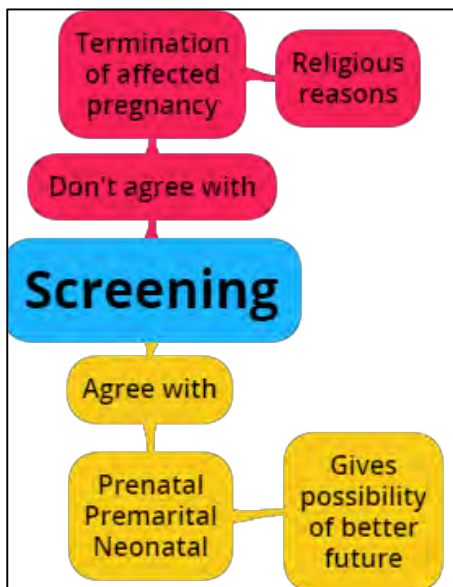


Figure 5.1: Summary of findings on attitudes towards screening

Chapter 6: Conclusions, limitations and recommendations

6.1 Conclusion

This study aimed to explore the experiences of parents with children affected by SCD in Cape Town, and discover how those experiences were shaped by knowledge of SCD and the perceived burdens of care, as well as assessing opportunities for improving genetic counselling services. In addition the study aimed to explore parent's attitude to preventive policies aimed at reducing the prevalence of SCD. Seventeen biological parents with a child affected with SCD were interviewed. Recordings were transcribed for thematic content analysis.

The level of SCD specific knowledge varied among the participants, ranging from no knowledge at all to being aware it is a genetic blood disorder. Many participants revealed a misunderstanding of mode of inheritance of SCD, with many basing their deductions of inheritance on prior knowledge of the disease. Prior knowledge also greatly influenced participants' expectations of life expectancy, although their expectations were not always concordant with the expectancies in SA.

It was found that participants experienced several burdens in raising an affected child. While some burdens had certain practical, everyday consequences, such as time and finance, others were more psychosocial in nature, such as lack of support as a result of immigration, and required managing on a more psychological level. The strategies employed by participants for this management were also explored, underlining certain strengths and weaknesses thereof.

It was found that the majority of participants supported all methods of screening for SCD. Even in instances where the participants indicated that they would not make use of specific services themselves, they agreed that it should be available to those who chose to utilise it, provided it remained the individual's choice. Lastly, while many participants had never received genetic counselling, benefits to those who had included increased knowledge and understanding of SCD and better acceptance thereof. There is a strong indication for the need of the improvement of genetic counselling services available to parents of children affected with SCD. The researcher feels that, ideally, parents of all SCD affected children should be seen by a genetic counsellor at least twice, once following the diagnosis and once, after a period of time has elapsed, to assess understanding and any psychosocial needs. Additionally, genetic counsellors should be available for follow-up referrals from doctors at all times. The genetic counselling services at RCWMCH

Haematology clinic is not currently this involved, mainly as a result of the few genetic counsellors available.

The research findings of this study have provided valuable insights, not only around the experiences of parents, but also about the genetic counselling services provided and their potential role. But the findings of this study cannot be generalised to all parents of children with SCD in SA, and as such more research is needed. It is hoped that the observations noted in this study, combined with the recommendations made will assist in providing more informed genetic services to those seen at the Haematology clinic. The researcher feels that this study could contribute towards improving understanding and subsequent care of individuals raising a child affected with Sickle Cell Disease.

6.2 Study limitations

- Due to limitations on the time available to conduct interviews and the time consuming nature of transcribing the interviews, analysis of the data was only started once all interviews were finished. This meant that any theories emerging from the first interviews could not be explored in subsequent interviews.
- The main purpose of this project was to provide the researcher with a learning experience to become skilled in qualitative research, which meant there was a distinct learning curve. Consequently the first few interviews conducted were not as good and insightful as the latter interviews, when the researcher had gained more experience.
- All interviews were conducted in English because of time and resource constraints. As this was not the primary language or mother tongue of most of the research participants it may have affected the quality of data gathered.
- Due to time constraints not many more interviews could be conducted. However, it was felt that saturation was reached as similar data was emerging from all interviews and the last few interviews did not lead to new insights.
- The purposeful sampling method used in this study has the potential to lead to some ascertainment bias, as the views and experiences of individuals who are unwilling to participate or less communicative are not included.

6.3 Practical implications and recommendations

Based on the findings of this study, the following recommendations are made. Firstly, improved communication and co-ordination between the Haematology and Genetic units at RCWMCH could be advantageous to ensure early referrals to genetic counselling services for parents with a newly diagnosed child or those who had never received counselling. In addition, follow-up appointments would be beneficial to parents who are not yet able to cope with their child's diagnosis. This would ensure all patients fully benefit from genetic counselling services.

Secondly, a simple fact sheet explaining the condition, its cause and inheritance should be given to all parents seen at RCWMCH Haematology clinic. Such a fact sheet is already available through the University of Cape Town Human Genetics website, and the routine distribution thereof may be helpful in correcting misconceptions as well as providing something that can be easily shared with family or referred to in future. The translation of this factsheet into other languages, such as French, could further improve the reach of this fact sheet. A similar factsheet, focusing on more medical aspects of SCD, is currently accessible at the clinic although not actively distributed. This fact sheet could also be distributed to primary health care professionals, ensuring all are well informed and aware of the condition.

It is also recommended that, although genetic counsellors routinely enquire about parents' prior knowledge of the disease in question, it might be prudent to pay extra attention to it in this group of patients, given that they often do have prior knowledge of SCD, possibly under a different name. It might be necessary to correct certain misunderstandings or explain some aspect in more detail as a result of what comes to light about prior knowledge.

It would also be very beneficial if both parents of an affected child are seen by a genetic counsellor once during their child's treatment at RCWMCH. This would guarantee both parents the chance to ask questions as well as ensuring information held by both parties are correct.

While genetic counselling services at RCWMCH are mainly aimed at parents, the young adolescent patients with SCD could equally benefit from it. As such, it might be valuable for doctors to also start referring their patients who are of a certain age for genetic counselling, instead of only referring parents.

The founding of a support group for parents of children affected with SCD could provide parents access to a much needed resource for social support. Exposure to others in the same situation could be advantageous by providing not only hope, but also a space where stories/advice/experiences can be shared.

Lastly, to ensure that parents and patients fully benefit from genetic counselling services, a checklist can be developed, to be implemented with every new diagnosis of SCD and marked of as completed. This list would serve as a reminder and could include a specific number of genetic counselling sessions at certain intervals, a session including both parents, child's genetic counselling session (if applicable) and a reminder to provide information sheet and details of support group.

6.4 Research recommendations

Further research is needed to quantify the level of knowledge parents have of SCD, how that knowledge is influenced by genetic counselling, and how well it is retained after a period of time. This will be more generalisable and give a clear indication of which areas of knowledge are most difficult to address. Conducting research into the experiences of parents of affected children in other South African settings would be also be valuable, as it might differ according to availability of services, the quality thereof or other variables.

An investigation into the knowledge of SCD among primary health care workers would assist in determining the need for increased education and advocacy regarding the diseases. These workers could often be involved in either correctly referring the patient when first presenting with symptoms or treating them during a pain crisis, and as such it is important that they are cognisant of the disorder.

Finally, it would be of great value to explore the knowledge and experiences of SCD among individuals who are affected themselves, possibly both as adolescents and adults, in order to identify the burdens they experience as a person affected with SCD living in SA.

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Appendices

Appendix I: Information sheet

Study title: Knowledge and experiences of parents with children affected by Sickle Cell Disease in Cape Town.

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Introduction and Summary

We would like to ask you to participate in a research study that aims to investigate the knowledge, experience and needs of parents caring for children affected with SCD. The study has been initiated through the Division of Human Genetics, University of Cape Town. The findings of this study will facilitate the identification of areas where health services available to parents of children with SCD could be improved, with a specific focus on the genetic counselling services that are offered. This study is the focus of a minor dissertation for completion of an MSc Genetic Counselling degree.

If you agree to participate I would like to talk to you about your experiences with caring for your child with SCD as well as your understanding of the disorder. The discussion may take up to an hour and will be recorded. The interview will be anonymised through the use of codes, but excerpts from the interview may be used in reporting on this study.

A possible risk to you will be the discussion of sensitive and emotional topics during the interview. If you become distressed during the interview you will be referred to a practiced Genetic Counsellor registered with the Health Professional Council of South Africa. Any actionable comments revealed during your interview will be acted upon.

While you will receive no immediate benefit from this study, long term benefit will be to, if necessary, improve the genetic counselling process and services provided.

Privacy

All information will be handled confidentially and everything you say will be kept private. Only the researcher will know your name and any discussion with the project supervisors will make use of an assigned study code only.

The interview will be audio recorded so that the researcher does not have to write much during the interview. The recording will be kept locked up until the research has been written up and will then be destroyed immediately. The data stored on the computer will have a code only and your name will not appear anywhere.

Access to the recorded interview as well as transcripts will be limited to the researcher, being kept either in a locked drawer or on a password protected computer. Information may be used for a thesis, publications in scientific journals and presentations at professional congresses, but names will not be included.

Additional Information

Your participation in this study is entirely voluntary. You can withdraw your participation at any point. The interview can be terminated at any time and the recording will then be destroyed. Withdrawing from the study will not in any way negatively affect your future access to medical and genetic services for your child.

Should you have any questions during the duration of this study regarding:

1. problems as a result of the research, or
2. questions regarding information about the project

please contact me or one of my supervisors (details at start of document).

If you have any questions about your rights as a research participant please contact Prof. M Blockman, Chair of the University of Cape Town Faculty of Health Sciences Human Research Ethics Committee on (021) 406-6496.

Familial, relationship and society

Child quality of life (school performance, school absences)

Stigma

Preventative policies

Premarital screening

Neonatal screening

Prenatal screening

Attitude to TOP and reason

Appendix III: Informed consent form

PARTICIPANT CONSENT FORM

1. I have been invited to participate in the above research project because I have a child affected by SCD.
 - 2.1 I have been provided with sufficient information and understand that the objective of this study is to investigate:
 - The knowledge and understanding of SCD among parents of affected children
 - The impact of having a child affected with SCD, and my experiences in this regard
 - The genetic counselling services provided to my child and my perception of it
 - The attitude among parents of SCD affected children toward preventative policies
 - 2.2 I understand that some of the questions may make me angry or sad. The researcher will refer me to a genetic counsellor if necessary. She will show me respect, acceptance and empathy during the interview.
- 3.1 I have been assured that all information will be handled confidentially.
- 3.2 I understand that the interview will be tape recorded so that the researcher does not have to write much during the interview.
4. I have been assured that the recorded and transcribed information discussed during the interview will only be made available to the researcher's supervisors with my study code number and that they do not know that it refers to my name.
5. I willingly agree to consent in taking part in the study and I have been informed that I may refuse to participate in this project and that I may stop participating at any stage.
6. has explained the information of the study to me in English. I am proficient in that language and my questions have been answered satisfactorily.
7. I understand that there will be no medical benefits to me from this study.

8. I have been assured that participation in this project will not lead to additional costs for me or my family and I will not benefit from it financially.

I HEREBY DECLARE THAT I HAVE VOLUNTARILY AGREED TO PARTICIPATE IN THE ABOVE RESEARCH STUDY AND THAT I AGREE TO HAVE MY INTERVIEW AUDIO RECORDED

Signed at..... on.....2015

.....

.....

Participant's signature

Witness

Appendix IV: Ethics approval documentation



UNIVERSITY OF CAPE TOWN
Faculty of Health Sciences
Human Research Ethics Committee



Room E52-24 Old Main Building
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16 January 2015

HREC REF: 905/2014

Prof A Wonkam
Human Genetics
IIDMM

Dear Prof Wonkam

PROJECT TITLE: KNOWLEDGE AND EXPERIENCES OF PARENTS WITH CHILDREN AFFECTED BY SICKLE CELL DISEASE IN CAPE TOWN (MSc candidate Katryn van Niekerk)

Thank you for submitting your study to the Faculty of Health Sciences Human Research Ethics Committee for review.

It is a pleasure to inform you that the HREC has **formally approved** the above-mentioned study.

Approval is granted for one year until the 30th January 2016.

Please submit a progress form, using the standardised Annual Report Form if the study continues beyond the approval period. Please submit a Standard Closure form if the study is completed the approval period.

(Forms can be found on our website: www.health.uct.ac.za/fhs/research/humanethics/forms)

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

We acknowledge that the MSc student, Katryn van Niekerk is also involved in the study.

Please quote the HREC REF in all your correspondence.

Yours sincerely

signature removed

PROFESSOR M BLOCKMAN
CHAIRPERSON, FHS HUMAN RESEARCH ETHICS COMMITTEE