Perspectives on Genetics Research and Cures for Sickle Cell Disease in Jamaica

by

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Thesis submitted in partial fulfillment of
the requirements for the degree of
Master of Science in the Duke Global Health Institute
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ABSTRACT

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Abstract

Background: Sickle cell disease (SCD) is the most common genetic disorder of the blood. Jamaica has the highest recorded SCD prevalence in the Caribbean (0.65%). SCD treatment options are limited though new genetics research technologies such as gene editing and gene therapy show great promise as SCD cures. An ethical framework based on social and cultural contexts must inform the conduct of genetics research and the introduction of these technologies to the Jamaican SCD population. Methods: This qualitative cross-sectional study used semi-structured in-depth interviews to investigate perspectives on genetics research and cures for SCD and the social, cultural and ethical factors related to these perspectives among 10 SCD healthcare providers, 10 SCD patients and 9 parents affiliated with the Sickle Cell Unit (SCU) in Kingston Jamaica. Results: Though they expressed some skepticism, participants were optimistic about genetics research. They believed it would lead to improved SCD treatment and advance SCD knowledge. Attitudes towards genetics research also pointed to potential ethical issues regarding autonomy, confidentiality and benefit sharing. Participants also viewed SCD cures positively, though 4 patients revealed they had no interest in receiving a SCD cure. Participants also described a number of social and cultural factors such as socioeconomic issues, stigma and information seeking behavior that contextualized these perspectives. Conclusions: We propose a framework that integrates these perspectives, and the social and cultural contexts to guide SCD genetics research and the introduction of gene-based

SCD cures in Jamaica. Researchers in genetics must work closely with the SCU to ensure that participants fully understand study aims and methods, and develop benefit-sharing models that will ensure that participants and the wider Jamaican SCD community benefit from research to which they contribute. As SCD gene-based technologies become more readily available in Jamaica in the long term, the local health system must prioritize related professional and public education and programming, as well as social and counselling services to adequately prepare patients and parents to receive these technologies.

Dedication

For my family, friends, mentors: thank you for your unwavering guidance and support.

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1. Introduction

Sickle cell disease (SCD) is the most common genetic disorder of the blood [2]. Global estimates state that over 300,000 children are born each year with SCD. Models suggest that this number could rise to 400,000 by 2050[3]. SCD originates in malaria endemic regions such as much of Africa, the Mediterranean Basin, parts of the Middle East and India, where the sickle hemoglobin (HbS) variant is believed to confer protection against the malaria parasite[4]. More than 80% of SCD births occur in Africa with much of the remainder spread across the African diaspora[3]. SCD prevalence in the Caribbean, for example, is second only to that of Africa[5]. Significantly, the first SCD case described in Western literature originated in the Caribbean[4,5]. Jamaica, where the coverage of newborn screening (NBS) programs exceeds 98%, has the highest recorded SCD prevalence in the region of 0.65%[5].

The sickle hemoglobin (HbS) is a structural variant of normal adult hemoglobin (HbA) caused by a mutation in the HBB gene [3,4,6]. SCD most commonly results from the homozygous inheritance of the $ß^S$ mutation, while other forms involve the inheritance of one $ß^S$ allele and one of a number of HBB mutations including ß-thalassemia and C[2-4,6]. The polymerization of the HbS largely defines the pathophysiology of the disease[2-4,6]. Hypoxic environments trigger HbS polymerization, leading to the sickling of the red blood cell, hemolysis and altered blood viscosity. The resulting occlusion of blood vessels, ischemia and inflammation characterize the disease's hallmark painful crises[3]. In these

events, acute pain affects bones mainly in the extremities, back and chest. Children may experience strokes as a result of ischemia and infarction, potentially leading to cognitive, motor and psychological impairments. Moreover, damage to the spleen during infancy also creates high risks of contracting life-threatening pneumonia, sepsis and meningitis in childhood[6]. Though acute events largely define the disease's pathophysiology, SCD is also associated with chronic complications like organ damage[2]. Repeated vaso-occlusion can result in parenchymal damage to almost every organ system in the body. This damage, particularly to the cardiopulmonary and renal systems, causes significant mortality and morbidity for patients beginning in the third decade of life[2,6].

The social and economic consequences of SCD are manifold. In the US, SCD disease related health expenditure is estimated at $1 billion[7]. Individuals living with the disease face psychosocial issues such as stigma, depression and loneliness, and poor quality of life[8,9]. Disease complications result in absenteeism from school and work, leading to widescale socioeconomic insecurity for patients and their families[3,10-12]. Moreover, the disease is associated with high rates of early childhood mortality and decreased life expectancy, particularly in low- and middle-income settings[3,6].

Globally, SCD has been historically neglected, though key organizations such as the World Health Organization (WHO) and the NIH have recently begun to recognize and address the global impact of the disease[3]. The WHO has committed to increasing international awareness, promoting equitable healthcare access, providing technical

support for disease prevention and management, and supporting SCD research[13]. However, while efforts such as newborn screening (NBS) and penicillin prophylaxis have improved health outcomes for those with SCD, life expectancy still falls decades short of the global average[6]. In many low- and middle-income settings, where the burden of disease is highest, many of these public health interventions remain widely inaccessible[3,5]. In the English and Spanish Speaking Caribbean, for example, NBS coverage barely exceeds 50%[5]. Moreover, effective and accessible treatment options remain limited. Up until 2017, hydroxyurea was the only approved pharmacological treatment for the disease[2]. More recently, a few others such as Endari have been approved by the FDA. Allogenic, hematopoietic stem cell transplantation (HSCT) remains the only approved SCD cure[2,4,6]. HSCT is successful in 85-95% of patients, with a 5-year event free survival rate of 91%[14]. The procedure, however, necessitates a suitable HLA-matched donor. Furthermore, the associated risk of infertility and endocrinopathies, and low accessibility of the procedure limits HSCT availability to only the most severely affected patients, generally in high income countries[4]. Therefore, the procedure remains mostly unavailable to SCD patients in low income settings.

Variation in symptoms and disease severity further complicates SCD management. Though the result of a single point mutation, SCD features significant phenotypic variation that belies its simple genetic basis[2,6]. Genetic and environmental factors, and their interaction are believed to play a role in SCD variability. The nature of

this, however, is not fully characterized. Research points to alpha-thalassemia coinheritance and genetic modifiers that influence the levels of fetal hemoglobin (HbF) as important genetic factors in this variability[2,15]. HbF, for example, serves a protective function as it impedes HbS polymerization[2,6,15]. Environmental factors are also believed to play a significant role in SCD phenotypic variation. Climatic and meteorologic factors such as cold weather, windspeeds, humidity, altitude, and air pollution, as well as socioeconomic factors have been shown to be strongly associated with disease severity[15].

Understanding the nature and mechanism of SCD variation could lead to more targeted and effective treatment and management. Genetics and genomics research have been essential in defining the nature and course of SCD throughout the disease's history. Advances in genetics and genomics research are now revolutionizing how we understand this disease complexity and how we approach SCD treatment and management. However, in low- and middle-income countries where SCD burden is highest, much of this research and emerging technologies remain inaccessible due to ill-equipped health infrastructures. Limited access to genetic tests, and sequencing and array technologies for example restricts genetics-related health data generation, and genetics research and analysis in these regions. As a result, available genetics-related health data are largely skewed towards higher income regions and European populations[16].

Recent advances also point to SCD gene-based cures such as gene editing and gene therapy as the future of SCD treatment[4,6,17]. These technologies alter the hemoglobin

composition of the red blood cell. Gene therapy achieves this by adding either a non-sickling ß-chain variant or a γ-chain globulin to produce HbF using viral vectors. Gene editing, on the other hand, repairs the β^S mutation or enhances endogenous γ-globulin expression by manipulating HbF silencing pathways[7,17]. Though these technologies show promising trial results, safety and efficacy issues remain[7,17]. Furthermore, trials have been conducted primarily in high income regions like the USA and Europe, biasing results towards these populations[4]. Finally, economic and infrastructural limitations will likely preclude treatment accessibility in low income settings where SCD burden is highest.

Genetics research and gene-based cures heavily rely on a foundation of patient, population, and disease specific health data to effectively inform disease management and treatment[16,18] That is, effectively treating and curing SCD patients in low resource settings will be enhanced by data from these patients. The Human Heredity and Health in Africa (H3Africa) initiative strives to ensure that health data from African populations help to inform disease management efforts in these populations. The initiative also works to build research capacity and develop the necessary infrastructure to facilitate genetics research in the African region[16]. This infrastructure includes biorepositories for biospecimen storage, as well as human resources. In so doing, H3Africa hopes to bring genomics and its related technologies to this population, focusing on a number of diseases including SCD[16]. Though little effort has been made to introduce such advances to the Caribbean, the region's high SCD prevalence makes it an ideal target for these technologies.

Moreover, treatments such as HSCT remain largely inaccessible for much of the region's relatively large SCD population. Similarly, gene-based cures will likely be available only to very few. Nonetheless, as more populations continue genetics research activities and move towards gene-based cures for SCD treatment and management, the Caribbean must be ensured to follow.

These activities raise a number of issues beyond accessibility and inclusion. As previously described, gene editing technologies continue to garner safety and efficacy concerns among SCD stakeholders[19]. Further, social and ethical concerns about genetics research and SCD gene-based cures have also emerged among these stakeholders. Therefore, issues regarding patient data generation, storage and use, as well as the social and cultural implications of SCD cures must be addressed. A few studies have engaged SCD communities in the US and Africa on these issues. They consider the social and ethical implications of genetics-based SCD research and intervention within these contexts. These studies identify issues such as dissatisfaction with current treatment options, fears about complications or side effects from gene editing and gene therapy, mistrust and a demand for transparency, concerns about cost and accessibility, and religion's role in health management and decision making[19-21]. No studies, however, have looked at individual and community perspectives on genetic research infrastructure like biorepositories and stored biospecimen use specifically for SCD research, or studied

attitudes towards both HSCT and gene based SCD cures. Finally, no studies have looked at these issues in Jamaica or the wider Caribbean.

1.1 Study Aims

Curative technologies like gene editing and gene therapy are the future of SCD management and treatment. To, successfully confront the SCD burden in high prevalence regions like Jamaica, efforts to support the generation of population specific health data through genetics and epidemiological research and the effective integration of novel technologies into patient care must be prioritized. While these efforts require the development of the local health and research infrastructure, they also require the development of region specific social and ethical frameworks. Therefore, research must be done to understand the ethical, social and cultural implications of these technologies within the Jamaican context. This study aims to fill the existing gap. The specific objectives of the present study are to:

1. Identify SCD stakeholder perspectives on SCD genetics research and related issues including biospecimen storage and use
2. Assess SCD stakeholder understanding of and interest in SCD cures
3. Identify the social and ethical implications of SCD stakeholder perspectives to help inform a potential framework for genetics research and SCD management

2. Methods

This qualitative, cross-sectional study engaged sickle cell disease (SCD) stakeholders in Jamaica on their perspectives on genetics research and cures for SCD. Ten (10) individuals living with sickle cell disease, 9 parents of children with sickle cell disease, and 10 health care providers were interviewed remotely one-on-one using the video conferencing platform, Zoom. Ethical approval was obtained from the Campus IRB at Duke University [2021-0014] and the Ethics Committee at the University of the West Indies, Mona [ECP 178, 19/20].

2.1 Setting

This study was set in Kingston, Jamaica but was conducted remotely over the video conferencing platform, Zoom. Participants were recruited from the Sickle Cell Unit (SCU). The SCU is part of the Caribbean Institute for Health Research of the University of the West Indies, Mona Campus in Jamaica. The SCU carries out research and clinical care functions involving patients with SCD. This specialist clinic is the only one of its kind on the island and serves patients and parents from all 14 parishes in the country.

2.2 Participants

Ten (10) SCD patients, 9 parents of pediatric SCD patients and 10 healthcare providers affiliated with the SCU were interviewed in this study. Eligible patients were those who were 15 years or older, able and willing to provide informed consent, and who were not acutely ill at the time of recruitment. Eligible parents were those who were 18

years or older and who had one or more children with SCD receiving care at the SCU. Eligible providers were doctors, nurses or social workers who worked at the SCU in clinical care or research. Providers were recruited through convenience sampling, while patients and parents were randomly selected from a list of patient information. A purposive sampling strategy was also used to ensure that male patients and parents were selected. Sample sizes were chosen with the aim of achieving thematic saturation within each group and across the entire study population.

2.3 Procedures

Interviews were conducted between September and December 2020. Random samples of patient and parent names were generated from patient files at the SCU. Eligible patients and parents were contacted by a research nurse employed to the SCU and affiliated with the study. A standardized script for personal contact was used to inform potential participants of the study and its aims and to arrange a time for the interview. Due to difficulties faced in recruiting male participants, a purposive sampling technique was used to ensure that male patients and parents were selected into the study. Thirteen (13) patients and parents that were enrolled were dropped from the study either due to problems with internet connection or failure to show up at the allotted time. Providers were recruited through convenience sampling. They were approached and, using a standardized script, informed of the study and its aims. A convenient interview time was

also identified. As a result, only those providers present at the SCU during the time of the study were likely to be recruited into the study.

Interviews were conducted remotely, using the videoconferencing platform, Zoom. A recurring zoom link was developed and sent out along with consent forms to enrolled participants. Consent forms can be found in Appendix A. At the time of the interview, participants were based in locations of their choosing. For parents and patients, this was typically their homes, while for providers this was generally the SCU. Participants were given the choice to have their cameras on or off during the interview. Each interview session began with the consenting process. While using Zoom's screen share functionality, consent forms were read and explained to participants over Zoom in the presence of a witness who was unaffiliated with the study. This was either another Duke University student or a nurse at the SCU. All participant questions regarding consent or the study in general were answered. The consent form was then signed by the interviewer with the participant's name and the date, and this copy of the consent form was emailed to the participant.

Following the consenting process, participants were engaged in one-on-one, semi-structured, in-depth interviews using a previously developed interview guide. Interviews were conducted in a mixture of English and Jamaican Creole. Interviews lasted between 45 and 90 minutes and were recorded using Zoom. A description of each interview was created to document any concerns and highlight any notable themes or ideas.

2.4 Data Collection Instruments

The instrument used in this study was a semi-structured in-depth interview guide. The guide was used to ensure that interviews were standardized and comparable. The guide was generally followed, and probing questions were asked at the interviewer's discretion. The interview guide adapted and modified the Sickle Cell Disease Genomics Network in Africa's (SickleGenAfrica) community engagement format and survey. Previously used in Accra, Ghana, Abuja, Kano and Lagos, Nigeri, and Dar es Salaam, Tanzania, these instruments were developed to explore specific issues in genetics research, including sample collection and storage in biorepositories. These instruments were modified into one document to fit the semi- structured, in-depth interview format, and to include questions that explored issues surrounding cures for sickle cell disease. Further questions explored knowledge of and attitudes towards SCD cures, individual experiences with sickle cell disease management and treatment, and the socioeconomic and psychosocial challenges related to SCD to better contextualize participant attitudes toward genetics research and SCD cures. The interview guide was modified and further developed throughout the data collection process to addresses challenges related to question clarity or comprehensibility. This interview guide can be found in Appendix A.

2.5 Analysis

Transcripts were automatically developed by Zoom software. The transcripts were then cleaned and edited for accuracy. Words or phrases in Jamaican Creole were rewritten

into Standard English, or otherwise clarified where it was determined necessary and where doing so would not interfere with the sentiments or ideas the participant sought to express. Participant demographic survey information was compiled into an Excel document. These documents were imported into NVivo 12 for analysis. Interview transcripts were assigned to cases and the demographic information assigned as case attributes. Transcripts were closely read and structurally coded based on interview questions and the general structure of the interview guide. Themes related to participants' perspectives on and attitudes towards genetics research and SCD cures, and relevant social, ethical and cultural issues were identified. These themes and corresponding codes were compiled into a master codebook that defined codes and specified their potential uses. Interview transcripts were then further coded based on these emergent themes. Memos were written based on each of the study's three aims to organize themes by aim and to summarize preliminary findings from this first pass thematic coding. Additional themes identified during these processes were added to the codebook and transcripts were recoded where necessary. Queries, including matrix coding and cross tabulation were then run to compare codes across participant types and to make comparisons between relevant codes. These queries were summarized in results tables and relevant and informative quotes were selected.

3. Results

A total of 29 interviews were conducted. While 42 participants were identified and contacted, 13 were dropped from the study, either due to failure to show up at the scheduled times, or insurmountable challenges with video call connection. Thematic saturation was achieved.

3.1 Participant Characteristics

Ten (10) Providers, 10 patients and 9 parents participated in this study. Participant age ranged from 22 to 64 years of age. Providers had the greatest average age of 44.6 years followed by parents (38.1 years) and patients (36.1 years). Approximately seventy-two percent of respondents were female. Only 2 male providers, 3 male patients and 3 male parents participated in the study. Approximately 82% of participants identified as Black and 93.1% identified as Christian. While most providers and patients (100% and 60% respectively) had some tertiary level education, most parents' (88.9%) highest education level was at the secondary level. Thirty percent (30%) of providers, 100% of patients and 55.6% of parents had a family member (other than themselves or their children) with SCD. Participants represented 9 of Jamaica's 14 parishes.

Providers were either doctors, nurses or social workers. Patients' reported genotypes that included SS, SC and Sß-Thalassemia, and a range of illness severities. Finally, parents included 6 mothers, 2 fathers, 1 grandmother, and 1 uncle who were either the child's primary caregiver, or co-caregiver.

Table 1: Participant Characteristics

	Provider	Patient	Parent	Total
	(N=10)	**(N=10)**	**(N=9)**	**(N=29)**
Age (in years)				
Mean (SD)	44.6 (14.2)	36.1 (10.9)	38.1 (5.80)	39.7 (11.3)
Median [Min, Max]	43.0 [29.0, 64.0]	35.0 [22.0, 55.0]	35.0 [31.0, 46.0]	35.0 [22.0, 64.0]
Self-Identified Race/Ethnicity				
Black	7 (70.0%)	8 (80.0%)	9 (100%)	24 (82.8%)
Mixed	3 (30.0%)	2 (20.0%)	0 (0%)	5 (17.2%)
Gender				
Female	8 (80.0%)	7 (70.0%)	6 (66.7%)	21 (72.4%)
Male	2 (20.0%)	3 (30.0%)	3 (33.3%)	8 (27.6%)
Highest Education Level				
Primary	0 (0%)	1 (10.0%)	0 (0%)	1 (3.4%)
Secondary	0 (0%)	3 (30.0%)	8 (88.9%)	11 (37.9%)
Tertiary	10 (100%)	6 (60.0%)	1 (11.1%)	17 (58.6%)
Employment				
Employed	10 (100%)	9 (90.0%)	5 (55.6%)	24 (82.8%)
Unemployed	0 (0%)	1 (10.0%)	4 (44.4%)	5 (17.2%)
Religion				
Christianity	9 (90.0%)	10 (100%)	8 (88.9%)	27 (93.1%)
Other	1 (10.0%)	0 (0%)	1 (11.1%)	2 (6.9%)
SCD in Family[1]				
Yes	3 (30.0%)	10 (100%)	5 (55.6%)	18 (62.1%)
No	7 (70.0%)	0 (0%)	4 (44.4%)	11 (37.9%)

[1] For parents: with the exception of child/children

3.2 Aim 1: Perspectives on Genetics Research

Excitement and Enthusiasm

Most participants spoke favorably of genetics research. They discussed why individuals with sickle cell disease might participate in genetics research and the importance of Jamaicans participating in genetics research. They saw it primarily as 1) the path to a cure or improved treatment options for sickle cell disease, and 2) a way to advance SCD knowledge. These positive attitudes are summarized in **Table 2**, and are explained in the sections that follow.

Table 2: Enthusiasm and Optimism Coding Matrix – Genetics research motivators by participant type

	Provider n=10	**Patient n=10**	**Parent n=9**	**Total n=29**
Better Options and a Better Life				
For Themselves	1	4	4	9
For Others	1	8	2	11
Advance Knowledge				
Medical	5	2	0	7
Personal	0	6	4	10

Better Options and a Better Life

Most participants viewed genetics research as a means to bring about or receive improved treatment for SCD (**Table 2**). Notably, many participants discussed genetics research participation in terms of receiving SCD cures. Parents and patients discussed advancements in SCD management and treatment that had led to increased lifespans for

those with the disease. They believed that genetics research would build on this work. One patient (Female, 30's) explained,

> "I'm just thinking that over the years medicine would have developed. So, I'm thinking that each year, or over the years, acquiring information or doing [genetics] research then, they would know exactly or of a type of treatment to help persons with sickle cell disease."

When asked why individuals living with sickle cell disease would participate in genetics research, most providers described the physical, mental and emotional challenges the disease created and the limits they felt it placed on patient's and parent's lives. They saw genetics research as way that patients and parents could overcome these challenges. One provider (Female, 40's) explained,

> "They wish to be quote, unquote normal. They want to be cured. They don't want to have a restriction hanging over them or…they don't want to be experiencing those complications anymore."

Similarly, another provider (Female, 50's) emphasized the emotional and mental burden created by the disease that caused many patients to desperately hope for the change they believed genetics research could provide.

> "The emotional and mental despair would really lead them to, to make that decision too. It's not just the physical, yeah. They have to just reach to a point where they say, you know, I just cannot live, I don't want to live like this anymore. I will, I'm willing to try something else."

Another provider (Female, 50's), speaking on behalf of parents of children with the disease, stated,

> "Parents… especially those who have had a lot of children with a lot of hospitalization… are anxious to find a way, you know, you don't want to see your child suffer, so you're anxious to find a way to cure to make things better."

Some patients and most parents of children living with SCD reaffirmed provider opinions. These participants emphasized their desire to participate in genetics research with the hopes of alleviating the challenges associated with their illness. Respondents also expressed dissatisfaction with current treatment options and their need to take medications each day to manage the disease. One patient (Female, 20's) explained,

> "Just taking these medications on every day. I will be willing, more than willing to be in this research. Because I don't like to be taking medications every day."

Patients believed that participating in genetics research would lead to new treatment options that minimized or eliminated the need for constant, long term medications entirely.

Altruism also appeared to strongly influence most patients' attitude towards genetics research (**Table 2**). This was particularly the case for those who reported less severe symptoms or challenges associated with the disease. Often, these patients had fewer or less painful crises and felt that the illness had not limited them in any way. When asked why she would participate in genetics research, one such patient (Female, 50's) with

mild SCD symptoms replied, "because for me, personally, it can benefit my family members and relatives and others in general."

For other who expressed altruistic motivations, while they recognized the limits the disease imposed on them, viewed their journeys with SCD with pride. When asked why she would participate in genetics research, a patient (Female, 40's) who was a self-described "proud sickle cell person with two teenagers" who was "healthy and… strong and hardly [had] crises", responded,

> "The fact that I'm helping others…as I said before, I love helping persons. And I don't want to see persons suffering as much as I did. So, for me to participate and to help others, I am willing to do that."

Altruism as a motivator also emerged among a few patients that seemed to accept the challenges associated with sickle cell disease and felt change would be too little, too late. This was particularly evident in one patient (Female, 30's) who, when asked why she would participate in genetics research replied,

> "Miss to tell you the truth I think a lot of, all right if I was younger, and I found out about this genetic research or this sickle cell cure, definitely I would try."

When probed further, the patient went on to say that though she believed genetics research would not benefit her personally, she saw its potential to help others with the disease. Of her willingness to participate, she explained, "I would love to see a change in it. Trust me, I would like to see people be free from this sickness."

The Advancement of Knowledge

To a lesser extent, the hope to advance or improve knowledge also related to participants' excitement and optimism towards genetics research (**Table 2**). This was particularly evident among health care providers who spoke of the potential for genetics research to advance medical knowledge about SCD and health in general. Some providers drew on their own experiences to discuss the importance of genetics research in generating SCD knowledge. One provider spoke about a recent study that sought to better understand SCD related pain. This provider (Male, 30's) highlighted genetic testing as an integral part of this investigation, stating,

> "I just recently submitted something that requests subjects to do genetic testing. And that's because… it was important to, it's very important to understand certain things about pain and so on."

Providers also pointed to the importance of Jamaicans with SCD participating in genetics research to create a wider and more inclusive knowledge base. In describing why she believed it was important for Jamaicans to participate in research, one provider (Female, 30's) explained,

> "If we can say this is our experience and this is what we found that works or this is how our patients are unique and we can contribute to that. And it might be that what is unique to us is also unique to another subsect of patients somewhere else. And it might be something that they're experiencing that's different from others in either the first world countries or places that are more temperate or whatever the differences that might be that are identified,

> we can at least, the more information that we collect and the more that we can actually study that information and try and find the differences and the similarities, is the more likely we are to be able to help whether sickle cell patients in general or we are able to tweak the, the management to help the different subsets."

Similarly, some patients reflected on the gaps present in SCD knowledge and saw genetics research as a way to fill these gaps. One patient (Male, 20's), for example, believed that though there was "way more now", he did not feel that "there [was] a lot of information out there" on sickle cell disease. The patient went on to assert, "the more information that can go, I think it would be better, just for general knowledge", and acknowledged the significant role of genetics research in achieving this goal.

More patients and parents of patients, however, saw genetics research as a way to advance personal knowledge about sickle cell disease (**Table 2**). More specifically, a few patients believed it offered a way for them to better understand their bodies and how to care for them. One patient (Female, 50's) explained that their desire to participate in genetics research was simply, "Because I want to know about myself, my body." Another patient (Male, 30's) explained,

> "I just probably would like to learn more or probably if there are certain type of food that I should have eaten or that I'm not eating right, or stuff like that. that would be the reason. I mean just about anything."

Parents and patients also believed that genetics research would offer insights into heredity. For one parent (Female, 40's), this meant tracing SCD throughout their ancestry, "to find out how far" back the disease went while for other parents and patients, this meant looking forward, to preventing SCD in future generations by allowing them to,

> "…know where it is coming from and… know who they're choosing. If that person has sickle cell, then you know that they cannot partner with that person because they will bring forth a sickle cell baby."

Skepticism and Reluctance

Participants also conveyed skepticism or reluctance towards genetics research. They discussed why individuals with SCD would choose not to participate in genetics research and how certain elements such as the type of sample requested factored into this reluctance. Participants' reluctance towards genetics research related to fears of invasiveness or loss of privacy, mistrust, fears of the unknown and the time and inconvenience involved with research participation. These deterrents are summarized in **Table 3** and explained in the sections that follow.

Table 3: Skepticism & Reluctance Coding Matrix- Genetics research deterrents by participant type

	Provider n=10	Patient n=10	Parent n=9	Total n=29
Invasiveness	5	7	7	19
Mistrust	7	4	3	14
The Unknown	3	2	2	7
Time & Inconvenience	4	1	3	8

Invasiveness

Fear of invasiveness or loss of privacy most powerfully influenced feelings of skepticism or reluctance towards genetics research. This emerged particularly among patients and parents (**Table 3**). Many providers discussed patient and parent reluctance to genetics research due to its perceived invasiveness. One provider stated,

> "You'll have the few that will tell you that they are, um the usual thing for anyone doing the research, they don't want to be a guinea pig right?" (H32F)

This provider suggested that some patients and parents would fear the testing, experimentation and exposure they associated with research. Respondents believed that DNA stored sensitive information and worried that, as DNA was unique to an individual, this information could be tied back to the participant. One provider (Female, 40's) built on this sentiment, explaining,

> "People don't want them to have access to their private information and they're not sure…what the researchers are going to do…your DNA is just your DNA. It's just one you. So, it will always be linked back to you."

Many patients and parents also expressed similar concerns about the intrusiveness of research. They viewed research as a probe into one's private life. As one parent (Female, 40's) bluntly put it,

> "People being people don't want people probing their private lives or whatsoever they're hiding. And so, people have many objections about people digging into their lives."

Patients and parents viewed genetics research as a process that would intrude on aspects of one's life that they wanted to keep private. These aspects included one's family history and details about one's health. Patients and parents worried that as DNA or genes contained information about both, genetics research could expose this sensitive information. When asked why some individuals would be reluctant to take part in genetics research, one patient (Female, 50's) for example explained, "I think some people are, don't want to talk to about their sickness. Don't ever and ever want to."

Most patients and parents, however, expressed fears about the physical invasiveness of genetics research. When asked why someone with SCD would choose not to participate in genetics research, one patient (Male, 20's) suggested, "it's just fear of a procedure, you, either, they probably think it's going to be painful or anything like that."

Concerns about invasiveness most often arose when participants were asked about the type of sample they would provide for genetics research. This was particularly evident among parents of younger children. One such parent (Female, 30's) stated decisively,

> "…if it's a case where there's a lot of needle poking and stuff and, and things like that, I'm not going to subject, my child to that. So, anything that has to, asks my child to go to any form of pain or stuff like that, I'm not going to subject my child to that. It would have to be a case where I have no other alternative."

Finally, one provider (Male, 30's) hinted at similar concerns when asked his opinion on prolonged sample storage. He explained,

> "Because the, if you're using it for secondary data, you're putting the patient at less risk because they don't need to approach them again to take another blood sample or hair sample, whatever It might be."

Mistrust and Fears of Misuse

For most providers, many patients and a few parents, skepticism and reluctance related to mistrust of researchers and fears of misuse (**Table 3**). Respondents described their own feelings of mistrust and those they heard among members of their communities. Providers expressed fears about the unethical use of genetic information collected for the purpose of research. One provider (Female, 40's) recalled and compared fertility research conducted in the 1980s to genetics research and worried genetics research could feature

the same "type of over enthusiasm that was dangerous to people's health". This provider explained,

> "I am always cautious about my over enthusiasm about genetic research and in sickle cell disease as well. Just, just from a, you know, be careful and, and a reminder perspective that that we aren't we aren't God, we are mere mortals."

Providers posited that while these instances of misuse were often the products of natural human curiosity or "scientific fervor", sometimes, this misuse stemmed from something "more malicious". Regardless of the source, fears of misuse, and mistrust greatly drove negative perceptions of genetics research.

Patients also expressed mistrust and fears of misuse. One patient (Female, 50's) worried that researchers would "[take] some of [her] genes" and use them for purposes beyond what they had initially stated. The patient emphasized,

> "You can't trust these people… Because suppose they're storing it and um, they go and use it, they pinch a piece of it for their own purpose."

One parent (Female, 30's) shared these concerns. She explained,

> "Sometimes they tell you that they're…collecting data for this purpose, and only to realize that it's not that."

For a few parents and patients, this mistrust was linked to religious beliefs. Of her reluctance towards genetics research, one parent (Female, 40's) expressed,

> "…the way how the world run now, sometime [I'm afraid you know to tell you the truth… because you know growing up they tell you about the mark of the beast and things that is going to happen or so and sometime you're afraid… Sometime, them people tricky, you know. Worse them want take over the world and want destroy Black races."

Respondents explained that misinformation or conspiracy theories spread by other members of their communities drove negative attitudes towards genetics research. In hypothesizing about why some may choose not to participate in genetics research, one patient (Male, 20's) suggested,

> "Maybe misinformation or wrong information will get across and some people have certain belief that basically, what they're doing is putting things inside of you. So maybe that would scare off some of them from participating."

Similarly, a provider (Female, 50's) offered,

> "You know, they believe that, you know, people have all kind of theories that you put some chip in them body, that you going control their mind, you know…all kinds of conspiracy things."

This provider went on to reference the community response to the COVID-19 pandemic, the efforts to develop vaccines against the virus, and the related vaccine hesitancy in defense of her position.

> "I mean, right now people saying they don't want take the vaccine and Bill Gates want to put something inside of them to track them and all kind of, I mean, so."

One patient (Female, 30's) similarly referenced the pandemic to speak about the spread of misinformation in the community, and how this could affect participants' attitudes on genetics research for SCD.

> "I don't quite remember but a lot of people were afraid of that injection. So, most likely when we talk about, what I heard the Prime Minister, the Prime Minister mentioned the other day that they are trying to find a cure for the COVID and a lot of people are bashing him about this 'Bill Gates' injection and all sorts of things so, people are getting views from various angles. Maybe the same thing might start when the sickle cell." (P35F)

One provider (Female, 40's) discussed the "ugly history of misuse" as a basis for some of these fears. The provider explained, "there are examples of these things being misused or used in ways that were not anticipated and not agreed to previously."

This provider explained further that these fears of misuse were valid and historically and contemporaneously founded and warned that "it was important to catch these" instances of misuse in future studies.

Fear of The Unknown

A few participants discussed a general fear of knowing or fear of the unknown as drivers for skepticism or reluctance towards genetics research. Respondents suggested that individuals "may object" to genetics research "if they…don't have enough information about it." When asked why some would choose not to participate in

genetics research, one parent (Female, 40's) stated succinctly, "People afraid of things that them don't know."

Time and Inconvenience

To a lesser extent, respondents cited the time commitment and inconveniences involved with genetics research as reasons for reluctance. Providers explained that those unreceptive to research may simply be those who either were unable to "devote the time and energy that's needed…for this type of genetic research" or were hindered by "where they [lived or] the distance."

One parent (Female, 40's) spoke of her reluctance to allow her child to participate in a long-term study, explaining of continued participation, "if it's the case that it's going back and forth with this, it doesn't make any sense."

Respondents conveyed hesitation towards research processes they deemed too involved or too great an inconvenience. As one parent (Female, 40's) summarized, "I guess some people can't be bothered with the whole heap of test, test, test, test (laughs). Some people don't like those things."

Finally, one provider reflected on the fact that, often, genetic studies tended to be inaccessible for Jamaican participants as many took place overseas. In such cases the ability to participate in genetics research became somewhat exclusive. As one provider (Male, 30's) explained,

> "They're going to have to do a lot of traveling and all of that is a huge thing to consider. So as far as I know, the persons

who have done it, have been persons who had you know living or monetary situation that they could accommodate all of that."

3.3 Aim 2: Perspectives on Cures for SCD

Participant knowledge of and experience with SCD cures and their attitudes towards SCD patients receiving SCD cures are described in the sections that follow.

Knowledge and Experience

Participant knowledge of and experience with SCD cures are described below. Eighty percent of patients had prior knowledge of at least one cure for sickle cell disease (SCD), while only 33% of parents reported some prior knowledge. These parents and patients tended to display more robust information seeking behaviors, obtaining knowledge about SCD from more sources (**Table 4**). However, some respondents knew little of the cures beyond their existence, including even their names. In explaining what she knew of SCD cures, one parent (Female, 30's) explained what she had seen in a Facebook video some time before. The parent offered the vague description, "But they did it, some family member of hers, some blood thing but they did it and it cured her."

Table 4: Patient and Parent Prior Knowledge of SCD Cures

	Patient n=10	**Parent n=9**	**Total n=29**
Know	8	3	11
Don't know	2	6	8
Average Number of Information Sources			
Know (SD)	2.62	2.67	2.65
Don't Know (SD)	1.50	1.83	1.67

Another patient (Female, 40's), seemingly alluding to bone marrow transplantation, asked, "so, what about the one that you can drain out the blood and get different blood?", when discussing knowledge of SCD cures.

Most respondents' knowledge of SCD cures was limited to bone marrow transplantation. However, many held misconceptions about the nature of the procedure. One parent (Female, 30's), for example, expressed confusion about the bone marrow donor and wondered whether the bone marrow would have to be obtained "from a dead person." Others simply expressed uncertainty about the details of the procedure and how it would be carried out. One parent (Female, 42) disclosed, "The procedure I don't understand. I really don't know about the procedure so. I don't understand how you go about that."

Many patients and parents explained that they had learned of these treatments from online sources including YouTube and Facebook. One patient (Female, 30's) confessed, "I only watched a video about that marrow transplant", before going on to admit, she did "not know much about it."

Others were introduced to bone marrow transplantation by friends or family, or healthcare providers. One patient (Female, 30's) explained,

> "Actually, I did hear somebody mention it a long time ago to me. Ask me if I did not have any sibling who doesn't have sickle cell disease. And if I do, he or she is that person for my cure…he actually he work over the hospital doing the x-ray, at the x-ray department."

However, one patient's (Female, 30's) experience with SCD cures was more direct. The patient revealed that she had been asked to take part in a gene therapy trial. The patient stated,

> "They got in contact with me a couple months ago to do the gene therapy. So, I'm looking into that. I started, I signed the consent form a couple weeks ago and started screening."

A few patients and most parents, however, had heard nothing about cures for sickle cell disease. One parent (Female, 40's) said that though she had seen things like bone marrow transplantation on television, she had never spoken about it in the context of sickle cell disease. One patient (Female 50's) even expressed skepticism about the existence of a cure. The patient explained,

> "From I was growing up, I hear there is no cure for sickle cell… I don't think there is a cure, I don't know…if somebody come and say, you know I'm cured, thank you, thank you Lord, I'm cured from it. Well, I'd have to see the results and see if that is so."

Providers shared their experiences with SCD cures, though many admitted that these were limited. Less than half knew of patients that had undergone these therapies, and often emphasized that these treatments were generally inaccessible for Jamaican patients. One provider (Female, 60's) spoke of a patient who "used to come [to the clinic] …as a young girl with her mom" and in "very, very early days [had] had a stroke." The

provider explained that the patient had gone abroad and undergone bone marrow transplantation and emphasized the positive impact it had had on her life.

Another Provider (Female, 40's) described a patient who had undergone bone marrow transplantation upon migrating to the US. She noted, however, the rarity of such an outcome for her Jamaican patients, explaining that, in this case, the patient was only able to access the procedure as part of a study.

> "Unfortunately, most of our patients can't afford it and it's either that it has to do with, I think she was a part of a study as well that she had gotten the procedure done through."

Attitudes Towards Patients Receiving Cures

All providers were excited and optimistic about cures for sickle cell disease (SCD). In particular, they viewed gene-based cures favorably and supported their patients receiving these treatments. All parents were open to their children receiving SCD cures, though half showed some hesitation. Patients, however, displayed more varied positions on SCD cures. While most patients were open to the treatment, either stating that they would receive them or that they would consider it provided that certain conditions were met, 4 said that they were not interested in receiving any of the SCD cures.

Participants' attitudes towards SCD cures related to 1) the belief that SCD cures would lead to a "normal life", 2) the recognition that SCD cures would not be "a magic pill" and 3) concerns about risks associated with SCD cures. These themes are summarized in **Figure 1** and explored below.

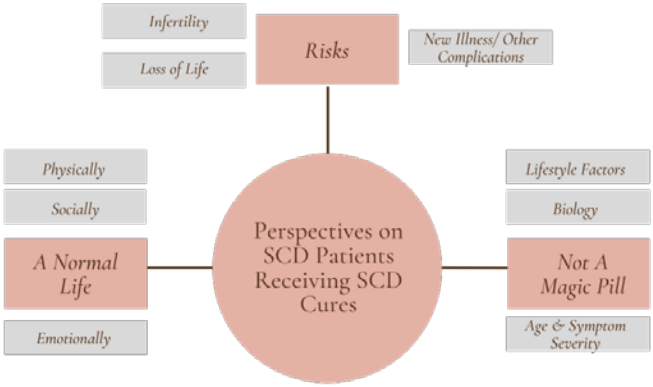

Figure 1:Factors Associated with Perspectives on Receiving SCD Cures

The Promise of a Normal Life

When overall attitudes towards cures for sickle cell disease (SCD), and perspectives on patients receiving SCD cures were considered, perhaps the most salient theme that emerged is that of 'a normal life' (**Figure 2**). Participants' perspectives were strongly associated with the belief that these cures were the keys to "a normal life" for patients. As one provider (Male, 30's) explained, "a cure is you being able to have that so-called normal life." When asked what impact she believed a cure would have on a patient's life, another provider (Female, 60's) ardently replied, "Oh, a positive impact! Do you know what it is to feel normal? It's a different feeling."

Figure 2: Word Cloud: Participants' attitudes towards SCD cures

Respondents conceptualized this idea of a normal life in terms of the physical, social and emotional impact they believed a cure would have on an individual's life. Respondents most often spoke of the physical changes that would result from receiving the cure. They discussed no longer "having to be in pain everyday", no longer needing to be "in the emergency room for pain", no longer worrying about "having jaundice" or your child having "this risk factor that can increase their chances of a stroke", but instead, having the "comfort that your child… [could] go on to lead a happy, healthy life, running around like other children." One provider (Female, 60's) explained,

> "It's a different feeling when you have a hemoglobin of seven. And all of a sudden you have a hemoglobin of 10,12. It's a different feeling. It's a different feeling when you wake up and realize that you are pain free…It's a different feeling when you can hit your foot on a bed post and don't have to worry about an ulcer."

Patients underscored these sentiments. One (Female, 30's) joked, "at least I would put on weight (laughs)" (P35F). Another (Female, 50's) exclaimed, "yeah, that would be so great. Oh Lord, thank you. No more joint pain at times!"

Respondents considered the social impact they believed the cure would have almost as often. They explained that the cure would remove the limitations that sickle cell disease had placed on patients. One provider (Female, 50's) explained that a cure would allow patients to "get a career, study, be gainfully employed." Patients would "be able to socialize even more", "go swimming anytime…go anywhere, any hour, anything" (H32F). Patients believed that a cure would probably "change how [they lived]" entirely.

One patient (Male, 20's) in particular, who described his symptoms as unpredictable and shared that "it just [attacked] constantly, one behind the other" seemingly without cause described the transformative effect he felt the cure would have on his life. He explained,

> "I have to live very cautiously from the elements, the weather, I would have to take care of myself. Keep warm, do this, do that. Basically, I could do what everyone else is doing. If there is a rainy time and I'm trapped outside I don't have to worry, I just walk come home, simple. Basically, it will just allow me to live life the way I wanted to."

Other patients and some parents echoed this view. They expressed that a cure would allow patients to do "what they [loved] to do". One parent (Male, 30's) explained that, for his child, this was swimming. He excitedly announced,

> "What you a say man? Right now, I know him would a say him want go back swimming you know. Right now, him would a say him want go swimming (laughs)."

To a lesser extent, respondents discussed the emotional effect they believed a SCD cure would have. According to a few parents and providers, this meant being free from worry, no longer having that "certain degree of awareness that they [had] a chronic illness that could later lead to complications", and no longer having to "[live] on the edge so much."

Those patients who already saw their lives as 'normal' saw no benefit to receiving these cures. This was evident among 3 patients who determined that they did not need to receive SCD cures. They revealed that they were not significantly bothered by SCD and were content to go on with life as it was. One patient (Female, 50's), when asked about receiving the cure explained, "Me? No, I don't think it bother me that much."

Another (Female, 50's) replied,

> "On me? At this stage in my life. No (laughs). No. a cure right now doesn't, I don't see how it benefits me. It's not like there's a limit, the average sickle cell person lives up to 63? And I don't think I'm average, so. I'm still looking at a good life. I don't really see how a cure, I don't think a cure would help me to live longer."

These patients did not feel limited by sickle cell disease and revealed that they faced no social or emotional challenges caused by the disease. They demonstrated strong

abilities to cope with the disease both physically and mentally. For one patient (Male, 20's) in particular, sickle cell disease was simply one facet of his life. The patient explained,

> "I'm great like even with sickle cell, I guess I'm comfortable enough with it, so I don't really have an issue with it. Like you know I think I've been coping fine."

This patient asserted that he lived "a pretty normal life" and believed he had an "awesome future ahead of [him]". Notably, among these patients, this was the only context in which the term "normal life" was used. None of these patients suggested that "a normal life" was something they lacked or needed to achieve. Rather, they implied that they already lived one. These patients also tended to have milder symptoms. They maintained that they "didn't have any severe issue" and reported that, on those occasions that they did experience crises, they were able to manage them effectively with pain killers or by simply, "[waiting] it out." The latter was the case for one such patient (Female, 50's) who revealed that she "[didn't] believe in this whole heap of medication" and instead, preferred to "rest, bear it [or] pray."

Not a Magic Pill

Though most wholly believed that SCD cures would have a large impact on patients' lives, some providers cautioned that a SCD cure was not a 'magic pill'. One provider (Male, 30's) remarked, "Magic pill? (Laughs) Um, I don't think that exists." Recognition of this fact appeared to slightly temper participants' enthusiasm about SCD cures. Another provider (Female, 50's) who initially stated that a cure would allow a

patient to "live a normal, as close to a normal life as possible", continued by asking, "although who lives a normal life? There's always something." When asked what sort of challenges could remain in a person's life after they receive a SCD cure, this provider explained, "So not necessarily that you get a cure, and your life just be fantastic. You may be left with a legacy of other issues." Overcoming the social limitations previously described, for example, were not likely to happen overnight. Patients would likely be left to struggle with the remnants disease's long-term effects. The provider reasoned,

> "Plus, the fact that you're maybe not, you may not be at a place professionally in your life that you can just pick up and start taking care of yourself, necessarily, if you have, if you haven't been going to school and you don't have no education. You know, just depends on the individual."

Respondents also discussed whether and which patients would benefit from bone marrow transplantation and gene-based cures. One provider (Female, 50's) asked,

> "Are you guaranteed to be free of the complications that…this disease has brought on you. Are you guaranteed that at the end of doing this gene therapy or whatever it is that you want to do that you will be free of all the problems?"

Participants acknowledged that SCD cures would be more beneficial to some SCD patients than others. Some respondents considered that age could minimize the benefits one could receive from SCD cures. One provider (Female, 40's) suggested that "younger

people might benefit more from this type of thing than, than older people." One parent (Male, 30's) similarly reasoned,

> "Well unless, unless they're at an age where or a stage where they feel like it makes no sense to them again (laughs) probably. Like you're 70 or 80 or 90 or going up and it doesn't make any sense to you again… but if you have a child that is ten, twelve, nine, five, and so… that would be good."

Providers argued that those patients who were older, or who had more severe SCD presentations were likely to have progressive damage and therefore would likely benefit less from SCD cures. One provider (Female, 40's) asserted

> "…older patients with, with more chronic problems… are hearing from us, you know, keep in mind that this type of therapy is to prevent progressive damage, it won't, it won't undo what is already damaged in the past"

Another Provider (Female, 40's) went on to posit,

> "…and I think that is why, in most of the studies that are doing DNA studies or stuff like that, they try to aim for the, a cure at an early age before damage is done to the rest of the body organs."

As one provider (Female, 40's) explained, those who received intervention early on would likely stand to benefit more as these treatments could prevent this damage and ensure that "they [wouldn't go] through, what some of the older patients [had] already gone through".

For other respondents, age related to acceptance. Those who grew somewhat accustomed to the challenges they faced for much of their lives were less driven to receive SCD cures. Of the gene-based cures, a 51-year-old patient who was not interested in receiving the cure stated, "for me, [I'm] old now, them not going change mine… right now, you go on live as you know." She implied that, as she was older, these cures offered little benefit to her. Instead, she considered the cures' benefits for her children with SCD. This patient explained, "Well, I'd like to know more about them to help maybe all my son or my daughter."

Another patient (Female, 30's) echoed this sentiment. However, in her case, she believed the cure would offer less benefit to her as she had passed the stage at which she could hope to have a child. For her, as this was the benefit she sought from a SCD cure, her age and life stage lessened the cure's perceived overall benefit. This was magnified by the fact that, like the previous patient, she had grown "used to" the disease and, and the pain was no longer "that bad like earlier."

A few others considered this issue in terms of an individual's physiology. This was most often tied to the experimental nature of gene-based cures. In this case, respondents considered the fact that patients could have little way of knowing whether and the extent to which the cure would benefit them in the ways that they had hoped. In discussing gene-based cures, one provider (Female, 40's) stated,

> "Right now, they're, they're not really treatment options because they're not approved so I always preface

> everything with this is this is experimental therapy. This is what we hope will happen, but I cannot say this with, with a particular degree of certainty that this is the case in anybody's bio physiology."

A patient (Female, 30's) shared similar concerns. She explained,

> "I would just love for it to actually benefit everyone because as I said we don't know how your body is going to react to something."

Finally, to build on this idea, one patient (Female, 20's) introduced the issue of lifestyle. She offered that a patient's lifestyle was likely to influence the benefit he or she received from these treatments. She framed her concerns around treatment trials as she wondered,

> "What if they then do the research on people who are sicklers, but their lifestyle is just party, party. They eat anything, any and everything and then you say there is an approval of this drug. And then you, you try on a different set of sticklers of sicklers who are just, they go to church, they go work, they come home, they eat their food, whatever it may be, and it doesn't work for them."

Risks

Concern about the risks associated with SCD cures also emerged as a significant factor from participants' attitudes towards SCD patients receiving these cures. While many providers maintained that bone marrow transplantation and the gene-based sickle cell disease cures were worth the potential risks involved, most participants expressed

concern about these risks. Providers said that they emphasized to their patients that these treatments were not risk free. One provider (Female, 50's) explained,

> "Is not like you just go do a bone marrow transplant like you just get some blood and then you're better. It has, you know, can have deleterious effects as well"

In terms of gene-based cures, one provider (Male, 30's) explained,

> "…at least in terms of the therapeutic ones so, gene therapy, it is high risk…I know the risks are improving and so on, but it is high risk."

Providers insisted that patients needed to understand the "possible adverse events" or "long term complications" associated with each treatment. They explained that patients also needed to consider the mental health challenges that could emerge from these procedures. Indeed, most patients and parents were cognizant of these issues and shared similar concerns. When asked what he wanted to know about gene-based cures in particular, one parent (Male, 30's) asked,

> "Will he still be able to live a normal life? Just basically that. Don't really want nothing to go affect him brain that all 'sheg' (mess) up all him schooling. Cause he is a brilliant youth right now."

Respondents worried that these treatments could have not only physical, but mental repercussions. Many parents feared that these potential side effects would outweigh the potential benefits of these cures. One parent (Female, 40's) asserted, "It

doesn't make sense you're going to get cure, a treatment that has a serious side effect." Another (Female, 30's) explained, "I'm not going to trade this prison for another prison one."

A few providers also voiced concerns about the impact that these treatments could have on fertility and reproduction. They believed that patients also needed to know how these cures would affect their offspring as one provider (Male, 30's) stated,

> "I believe, I believe the bone marrow transplants definitely result in um infertility and so on. So, you know, even, even when they're successful, so they're definitely negative outcomes, and they do need to take all of that into consideration when they are deciding what they're going to do."

A few respondents expressed fears about particular aspects of the treatments. For most, these fears stemmed from things they had heard or that they had seen online. One patient (Female, 30's) described a video she had watched in which a young woman with sickle cell disease had undergone bone marrow transplantation. She detailed some of the side effects the woman had experienced and expressed that "[she] was a bit afraid of the hair loss and the tongue swelling." Another patient (Female, 40's) said of the treatment that she had "heard that it [was] 50/50." That is, there was an equal chance that the procedure would be successful as unsuccessful. Moreover, as one parent (Female, 30's) explained, "you [could] go to through the surgery and you [could] die."

One patient (Female, 40's) emphatically rejected the idea of undergoing chemotherapy. She stated, "normally 99% of persons who do chemotherapy always die" and declared that she didn't "want no chemotherapy". Another's (Male, 20's) concerns lay with the HIV-derived viral vector used in gene transfer therapy. The patient explained, "they'll give you a mild form of HIV, that won't spread or something like that but I'm writing off that one as well."

A few patients and parents also expressed concerns for bone marrow donors. They wondered about the potential impact that donating bone marrow could have on their loved ones. One patient (Female, 30's) asked, "What will happen to my siblings…who have given me his marrow transfusion? What would happen to him?" A parent (Female, 30's) similarly wondered, "So, when they give their bone marrow, it doesn't affect them?"

Notably, some patients and parents expressed worry about a somewhat less tangible risk. These worries were specific to the gene-based cures that would alter parts of the DNA. They feared that altering their DNA, or that of their children, would erase the genetic link between them and members of their biological family. One parent (Female, 40's) asked, "So changing his genes would mean that, what, he's not a DNA match for his father or me?"

Few others worried that these processes would fundamentally alter their identities. These respondents believed that "all of us are born with unique genes" and that "changing a person's DNA would mean that you're changing who they really are." One

parent (Female, 30's) reasoned, "If you change their DNA that means they're no longer themselves. They are somebody else." Notably, these views, though rare, were more common among parents.

3.3 Aim 3: Ethical, Social and Cultural Issues

From participants' attitudes towards genetics research and cures for sickle cell disease, as well as their overall experiences with and perceptions of life with SCD, a number of ethical, social and cultural issues emerged. These issues are explored in the sections below.

Ethical Issues

Participants' attitudes towards genetics pointed to three main ethical issues. These issues included autonomy, benefit sharing, and confidentiality. They appeared to relate to participants' concerns about participating in genetics research, particularly those about invasiveness and loss of privacy, mistrust, and their hopes of obtaining better treatment options for and knowledge about SCD through research.

Autonomy

Respondents discussed their views on consent for the use of samples donated and stored for the purpose of genetics research. These discussions focused primarily on where and how long samples could be stored, and who could use donated samples and in what ways. They also included perspectives on ownership and control of donated samples. Participants' views towards autonomy and control pointed towards a preference for one of three types of consent: 1) specific consent, 2) dynamic consent, 3) broad consent. These types of consent are defined in **Box 1.**

Box 1: Types of Consent for the Use of Human Biological Samples[1]

Specific Consent: Allows the use of biospecimens for an immediate specific study or purpose in a specific, previously defined time. All uses of the sample must be explicitly outlined. Does not allow the use of biospecimens in future research or undefined research

Dynamic Consent: Requires constant communication with donors to obtain permission for the use of biospecimens in studies on a case-by-case basis

Broad Consent: Allows the use of biospecimens for an immediate specific study or purpose and undefined future study at any time with certain restrictions and governance

When attitudes were considered across all respondents, no unanimous position on autonomy emerged. While half of the participants favored stricter sample control through specific or dynamic consent, half believed this control was unnecessary and felt that broad consent was sufficient (**Table 5**). However, some differences were evident across participant subcategories. For patients and providers, these differences aligned with the extent to which each participant group expressed concerns about mistrust and misuse in research. Therefore, most providers, who spoke largely about mistrust and misuse, leaned towards stricter control. Most patients, on the other hand who discussed mistrust less frequently tended to favor looser control. Parents, however, were more representative of the entire study sample with half favoring one position and half favoring the other.

Table 5: Preferred Form of Consent, by participant type

	Provider n=10	**Patient n=10**	**Parent n=9**	**Total n=29**
Broad	2	7	4	13
Dynamic	6	1	4	11
Specific	2	1	0	3

Most providers believed researchers were to seek dynamic or specific consent for the use of stored samples. One provider (Female, 40's) who favored dynamic consent explained,

> "I…personally believe that you know if samples are being stored for long term…Then once, once somebody wants to go into those stored samples, then patients should be reconsented at that time to allow for these samples to be used in whatever way they're being proposed to be used."

Another (Female, 50's) who appeared to favor specific consent maintained that researchers were to inform participants of the precise study details at the time samples were collected. This provider believed this would allow participants to be both willing and able to make informed decisions about research participation. She emphasized the need for "everything [to be] defined to the T", and argued that "if it's too open ended, people [were] not going to want to give it because they [didn't] know where it [was] going [to] turn up".

Providers also discussed a similar need for total transparency and specific consent in relation to sample storage. One provider (Male, 30's) asserted that donors "definitely should know how long the sample [was] going to be stored" at the time of sample donation.

Providers reasoned that donors retained ownership over their sample. When directly asked whether a donor should have control over the sample, one (Female, 50's) pointedly stated, "Yeah. Well, I mean, you know, it's yours."

As such, providers held that donors had the right to dictate how their samples were used. One provider (Female, 50's) suggested that facilities or organizations like biobanks were stewards of samples donated for a specific intent, "entrusted to make sure that it [was] used for that purpose only." Some providers also suggested that control extended to the ability to determine when samples were to be destroyed or discarded. One provider (Male, 30's) stated, "I think that they should be able to request that it's removed or destroyed when they want it to be."

Those who favored dynamic consent particularly, believed that researchers were to contact participants periodically to determine whether they wanted their samples to be discarded. When asked about limits for sample storage, one provider (Female, 50's) mused,

> "Maybe it is that we should contact patients or stay in contact with patients if they change their view about in 20 years (laughs). You know, maybe we can have time periods, you know. Do they still feel that way? People change."

However, 2 providers spoke in favor of broad consent as opposed to the dynamic or specific consent described above. One Provider (Female, 40's) maintained, "I mean, I don't think…the patients should have a say in that once the general consent was given before." This seemed to suggest that donor control ended the moment the sample was signed over or, as another provider (Female, 20's) put it, "the moment they [had] given the right for it to be used for other research purposes."

Similarly, most patients and half of the parents favored broad consent. These respondents who worried less frequently about misuse and misconduct in research, believed that specific control over donated samples was unnecessary. As such, they allowed, these samples could be used for any purpose researchers saw fit, with some restriction or governance. One patient (Female, 40's) explained,

> "If I agree to a research, and you want to collect some samples, after you collect the samples, then I don't need to have any control over what you do with it."

Similarly, when asked whether she wanted control over or a say in what happened with her child's samples, one parent (Female, 30's) responded, "No…I don't see where that is necessary."

These respondents appeared to dissociate the sample from its donor. Unlike providers, they did not perceive samples as the donor's property after it had been donated. It was not tied to the donor in any meaningful way. Emphasizing this fact, one parent (Female, 40's) asked, "if the blood leave him body, what could he, what…it going to do it? What it going to do to him when it gone out of him body?" As the sample had already left her child's body, she felt it no longer had any real bearing on his life and as such, could do no real harm. Therefore, she had no reason to seek control.

Other respondents appeared to dismiss this control as not only unnecessary, but impractical. One patient (Male, 30's) explained that having control over donated samples was of "no use to [him] because it's not like [he knew] what to do with it", while one

parent (Male, 30's) announced, "it's not like I'm going to be the one…testing the sample. So, it doesn't make any difference or make any sense to me."

Notably, a few others believed and accepted that having control over the donated samples was impossible. Their leaning towards broad consent did not simply stem from their not wanting to have control. Rather, they believed that once donated, the sample was beyond their grasp, free for researchers to use as they saw fit. One parent (Female, 30's) asked, "If I give it for one reason and they want to use if for another reason, who am I to say?" This sense of powerlessness emerged quite frequently among patients and parents. One such patient (Male, 30's) captured this sentiment when he explained, "I couldn't stop them 'cause I don't think I would have that privilege" when asked how he felt about other groups or organizations using his donated samples for other work.

To a lesser extent, some patients and parents relinquished control of their samples on the condition that researchers used them for a purpose they deemed acceptable. These respondents also tended to rely on researcher reliability and truthfulness. They were willing to trust that these researchers were honest about their intentions and were unlikely to cause harm. Therefore, they could allow looser control than those participants that worried about misuse or misconduct. One patient (Female, 40's) hoped to limit sample use to sickle cell disease research, stating,

> "That's not my concern, I don't know, what you, I don't, once it has to do with sickle cell and you wanted to do, I don't business what you want to do. That's not my concern anymore… You come and you say, can I have your sample,

> we're going to do whatever, once you're being truthful and you're doing what you said you wanted to do, then that's fine by me."

Another patient (Female, 30's) stretched these limits to simply include research of any kind, explaining, "as long as it's not a case where they're using it to do something negative. So, if it's for research purposes then, no. It's okay with me."

Half of the parents and few patients, however reported their desire to have stronger control over their children's samples through dynamic consent or specific consent. In defense of dynamic consent, one parent (Female, 30's) maintained, "if you want my child's sample to do something, I think it's only fair for you to ask for my permission"

This parent expressed the strong need to know exactly how the child's sample would be used in order to make an informed decision about research participation. She asserted,

> "For me, it's, it's transparency, because if, if you are going to say, okay, I'm going to do A and I see where A is looking more like B then I won't want to participate. So, a lot of times people don't want to go through those things because they are not being upfront and straight with them. They are not telling you, they're not saying everything."

Confidentiality

Participants also talked about the need for confidentiality in genetics research. As previously described, many respondents expressed concerns about the invasiveness of

research and prioritized confidentiality and privacy. Some seemed to consider it most important when deciding to participate in genetics research. When asked specifically what rights donors should have over their donated samples, one provider (Male, 30's) responded,

> "I think that they should definitely, should be deidentified and it should be private. So definitely, their name should not be on it."

One patient (Male, 30's), when asked the same question, similarly responded,

> "As long as information is going to keep strictly confidential, is not like it's going to be something that's bouncing around."

Confidentiality also emerged among respondents when they discussed their concerns about genetics research. They emphasized the need for researchers to ensure that patients were deidentified and that there was no linkage between them and their genetic information. One patient (Female. 50's) suggested that she would have no concerns about researchers using her genetic information "once [she knew] it [was] going to remain confidential."

One provider (Female, 60's) took this discussion further. She considered the possible risks, were a participant's genetic information to be exposed. She talked about the potential for this information to fall into the hands of insurance companies, leading to "some legal and…personal ramifications" for the individual. She explained,

> "Like if a disease hasn't been manifested yet… you have the gene for say breast cancer mutation or some other type of illness that may come on later in life. Then that could be detrimental to the person's access to insurance."

As previously stated, some participants seemed to believe confidentiality was most important in genetics research. For some participants, their acceptance of the notion that donors did not need to have direct control over their samples was contingent on confidentiality being maintained. One patient (Female, 30's), for example, when asked about sample use stated,

> "…it's not a case where they definitely know that they're using (participant's name) sample and that with most research or majority it's like a number I think is linked to the link to the individual, to the patient. So, it's not a case where it's like personal information is being exposed. So, there's not much concern there either."

This patient explained that she had no concerns about her samples being used by different researchers or organizations, as she felt that confidentiality or the inability to connect her to these samples was enough. These perspectives recalled respondents perceived disconnect between donors and their samples. Confidentiality cemented this disconnect. One parent (Female, 30's) underscored this by explaining, "if they want to misuse it as long as it's not going to lead back to anything with my child, they can do what they want to do with it."

In other words, confidentiality would ensure that any ties her child could have with the sample had been definitively severed once it had been donated. Moreover, maintaining donor confidentiality would limit researchers' capacity to cause harm. As such, this parent needed no say in how the sample would be used or how it would be stored.

A patient (Female, 50's) endorsed this position, though she framed the argument somewhat differently. Rather, this patient explained that the extent to which she insisted on being detached from her sample depended on how it would be used. She explained,

> "If my sample is taken and they said that they're going to use it for sickle cell research. And then down the road now, they, they, somebody choose to use it to do some research to manufacture some cosmetic product or, so I am not with that. And I wouldn't want my name also to be associated. It all depends on what it's being used for as I said… It depends on what their going to use sample for you understand and might not want my name to exposed. I'm a private person."

Benefit Sharing

Respondents emphasized the importance of benefit sharing in genetics research. Excitement about genetics research often stemmed from its perceived benefit. Therefore, while there was some disagreement regarding the nature of this benefit, participants tended to prioritize awarding these benefits to those who participated in the study (**Table 6**). Providers, for example emphasized the need for those who made contributions to a given study to receive some benefit for that participation. As described previously, some

participants viewed the return of results as the benefit, in which researchers would share the information obtained through the study with its participants. One provider (Male, 30's) implied that it was important that participants were made aware of the advances in SCD knowledge and management that resulted from the research, and that they knew "that they actually played a role in this new development". Another (Female, 30's) similarly stated, "Every research that we do the patients must get an outcome of the results. They must be informed."

Table 6: Who Should Benefit From Research, by participant type

	Provider n=10	**Patient n=10**	**Parent n=9**	**Total n=29**
Community Benefit	2	4	2	8
Participant Benefit	3	4	4	11
No Benefit	0	3	3	9

Patients also spoke of their desire to know the results of research. One patient (Female, 50's) who had previously taken part in a study, spoke of her discontentment with not being appraised of its results. She complained,

> "That is what I don't like. At least come and say you know the, what we found was that or that, yeah. But we never got back any information."

Other respondents considered benefits in terms of a concrete product of research, in this case, novel treatments for SCD. A few parents hoped that if research their children

participated in yielded new SCD treatments, these children would benefit from these treatments. One parent (Female, 30's) hoped,

> "If they find a cure or some way to decrease the, the sickle cell in your body or something, they [could] just count her in."

Similarly, one patient (Male, 20's) believed,

> "Basically, who took part in it, basically, to me, should benefit first because one they've been suffering a lot lately, and if they can do anything to help them, such as lessen their pains or lessen their worries, I think they should be the one that would benefit first."

Many providers shared a similar stance. One (Female, 50's) hinted at the structural challenges Jamaican patients faced that hindered their access to healthcare. She believed that researchers held an obligation to grant access to those who participated in, and thereby contributed to the success of these studies. This provider stated,

> "So, you have helped this medication to be put out on the market, but then you don't have any access and you can't afford it. I don't think that's right (laughs). When your blood, your, whole participation in the research helped to put it out there. I think that these patients should at least have access or help or, or they should be helped to, to be able to afford access, whether its financial or otherwise to the product."

Some respondents, however, allowed that only those studies that yielded benefits to the researchers should provide benefit to the participants. One parent (Female, 30's) stated,

> "If it's a case where that sample is used, and they got benefit from it. It's only fair for the person that it comes from get benefits also. But if it's a case, where it's a free, then it's okay."

And a few patients and parents believed that, as research participation was voluntary, participants were not "entitled to anything". One parent (Male, 30's) explained,

> "There's some things in life, you can't get benefit from it when it's like for the good of other people…some people need help and if you can help someone its best you just do it."

To a slightly lesser extent, respondents proposed that the wider community was to benefit from research. One provider (Male, 30's) maintained that while individual participants were not to receive direct compensation from research, they, along with the wider community would indirectly benefit from "certain discoveries" that "[came] out of research." Another provider (Female, 60's) referred to Jamaica's large sickle cell disease population and described her hope that, "at the end that there'll be benefits. When I say benefits, I'm hoping that many persons can benefit from it." This provider wondered,

> "So, for example, if persons are part, to benefit from…any sort of you know, genetic research, once that is over and if proven to be successful, then how does that translate to helping our patients here who suffer from sickle cell

> disease?… So, the benefit as you asked, would be the fact that we have a large sickle cell population here in Jamaica, that we have patients to be able to do this research. However, when that is finished. How does the other, the others who have sickle cell disease benefit hear in Jamaica?"

Patients and parents also expressed their belief that the larger SCD community was to also benefit from the tangible products of research. One patient (Male, 30's), when probed further, also specified, "whoever needs the help first or, if I should range it, then I say who needs it the most should benefit first."

One patient (Male, 30's) however suggested that, as doctors and researchers were to be compensated for their efforts, any emerging treatments or technologies were to "come with a cost" for those who did not directly contribute to the study.

Social Issues

A number of social or structural issues emerged from participants' perspectives on genetics research and cures for sickle cell disease, and from their descriptions of their overall experiences with SCD. These issues are broadly categorized under socioeconomic challenges and stigma and are described below.

Socioeconomic Challenges

Of the challenges faced by those living with sickle cell disease, socioeconomic difficulties were among the most salient. Respondents often described socioeconomic challenges in relation to SCD treatment, cure access, and the healthcare access respondents hoped genetics research, and research in general could provide. These

challenges also emerged through discussions about the educational, professional and financial hardships caused by SCD.

A cyclical socioeconomic framework was developed based on participants' description of these issues and is shown in **Figure 3**. Education appeared to relate to employment, employment appeared to relate to income and access to healthcare, and both income and access to healthcare appeared to relate to education. Each element of this framework is explained below.

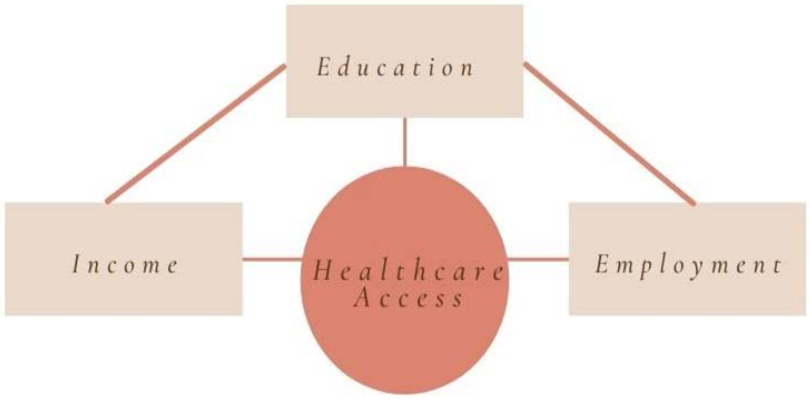

Figure 3: Socioeconomic Framework

Healthcare Access

Challenges associated with healthcare access emerged from participants' discussion of SCD cures. Respondents worried that Jamaican patients would be unable to benefit from cures for SCD. For most, cost was the prohibiting factor. One provider (Female, 40's) explained that "the initial numbers…out of some of the gene therapy trials

[were] astronomical amounts of money" that Jamaican patients were unlikely able to afford. Particularly given an exchange rate of "$150 to one", providers explained, the cost of these procedures would be "very prohibitive." One provider (Female, 30's) emphasized that cost was "already an issue with just normal treatment or normal management," suggesting that, with these newer gene-based technologies, this issue would only be magnified. Most providers also believed that Jamaica lacked the necessary infrastructure to administer these cures. One Provider (Female, 40's) explained, "When you hear of the cost associated with it and stuff like that, I don't think…our health system can manage that whether private or otherwise."

Providers cited the fact that bone marrow transplantation, though approved for general use, was still inaccessible for most Jamaican patients, due not only to its cost, but also to the fact that it was not offered on the island. To receive the treatment they explained, patients needed to "fly out to the states or to another country to do it" which introduced another set of "financial and logistical challenges."

Patients and parents also expressed concerns about the cost of these treatments. One parent (Female, 30's) explained of bone marrow transplantation, "From what I heard about it, I know that it's pretty expensive. And access to it now would definitely be a no, no."

One patient (Female, 30's) similarly worried that bone marrow transplantation "would be costing" and that "she probably [would] have to go overseas" to access it, suggesting it was not a treatment that was currently available to her.

However, these financial and infrastructural constrains were not only limited to SCD cures. They extended to customary diagnostic, preventative and mitigating SCD treatments. When asked about their experiences with their SCD medications and their ability to keep up with them, most respondents reported that financial limitations were among the main barriers to treatment adherence. Providers explained that patients would often tell them that, "they would like to take medication, but they [didn't] have any at home." Providers also reflected on the fact that, due to these challenges, patients were often "not able to access the optimal care that they needed" or to even buy necessary medication "because of lack of funds." Patients then, one provider (Female, 60's) explained, often experienced adverse health effects as a result, returning to the clinic or hospital "either in pain or [after] the infection [had] gotten worse". Corroborating this account, one patient (Female, 30's) explained that she was forced to purchase the cheaper brands to manage her disease, though often finding that "the cheaper brand, sometimes…[didn't] work as much as the other one would."

Some patients or parents were unable to run the tests required to make a diagnosis, monitor a health concern or better characterize their condition. One parent (Female, 30's) revealed that providers had found an abnormality in her child's brain and spinal cord. To

better understand and characterize this abnormality, it required extensive testing and imaging. The parent explained,

> "They were doing MRI, a lot of MRIs, MRIs, MRIs. And it hasn't been moving and…it was very costly. And they referred her up by UWI at the UWI clinic and I was going there, going there. And they're doing MRIs and…it's not moving, so I just let her stop by myself because it's costing me, even though it's her health, and I can't afford it. MRIs are very expensive."

Respondents often spoke of financial barriers in cases related to hydroxyurea. Providers explained that the treatment was costly and required "a lot of blood tests and…more monitoring." One provider (Female, 30's) explained,

> "There are quite a few patients that we would like to offer like hydroxyurea treatment option to but unfortunately because they can't afford the test or the medications, then, patients might be reluctant to try."

Another provider (Male, 30's) further stated,

> "I have persons who want to get on hydroxyurea and are trying to find ways of affording it. So, they might come and ask how they could fund it."

For some patients and parents, the structure of the health care system compounded these challenges. Respondents explained that as the SCU was not a part of the public health care system, its patients were not able to benefit from the free or discounted tests or medications offered through public labs or government pharmacies.

Instead, they had to pay for these services at the University hospital or at private labs. One Provider (Female, 40's) explained,

> "Any blood tests that we request, it has to usually go through the University Hospital. Which you have to pay to get done or it has to go through a private lab, which of course is paid to get done."

Similarly, one patient (Female, 50's) complained,

> "I have a issue too with, um the government health system where they change it, where you have to have a prescription from a government health center to, to get your prescription filled at the government pharmacy. Now, I think this is unfair to sickle cell persons…because the sickle cell clinic is not funded by government. So therefore, their prescriptions are not honored by the government center."

Providers went on to explain that there were sometimes "ways around things" that allowed patients to benefit from these public systems. Providers could, for example, "refer them through the public health care system" to receive these tests for free or at discounted rates. Moreover, patients could take their prescriptions to the Children's Hospital or the Kingston Public Hospital where doctors could rewrite them. However, these systems were sometimes limited in the type and quality of care they could provide. One Provider (Female, 40's) explained,

> "Not all of the tests that are available in the public health care system… And sometimes even when we refer them through the public health care system, sometimes there's a delay in getting back the test in time for the clinical follow

> up. And so, we end up where patients are kind of lagging with their treatment."

Furthermore, as one patient (Female, 50's) explained, if patients chose to go through the public health care systems, they would have to "sit down the entire day, just to get a government prescription" while sometimes in severe pain.

Limitations in the healthcare system also extended to the overall availability of medicines or equipment essential to SCD care. Some patients and parents reported that they faced difficulty acquiring medications like morphine and hydroxyurea from local pharmacies. One parent (Male, 30's) explained that when he first received his child's prescription for hydroxyurea, "all the pharmacies [he] contacted said that they didn't do it."

Many of these limitations also appeared to disproportionally affect those from rural parts of the island. One provider (Male, 30's) described what he observed as the disproportionate number of children in the rural regions who had had a stroke. Though he admitted that he lacked the data needed to verify this observation, he explained, "Children that I've seen in country clinic, they tend to have had a stroke, more than the ones that I've seen in Kingston." He hypothesized that this may have been due to accessibility. He explained,

> "And there's only one transcranial doppler machine in Jamaica. And even though the sickle cell unit is, you know, a central place where people can come, there are sickle persons and sickle cell children all across the island. So, a

> lot of people are not getting transcranial doppler and getting that you know assessment."

This resulted in the lack of proper diagnosis and preventative care, possibly leading to poorer health outcomes for children in these rural regions.

Notably, for many respondents, study participation was, therefore, the only way that individuals living with sickle cell disease could access care, and both established and novel SCD treatments. One parent (Female, 30's) explained that her daughter's affiliation with a study on hydroxyurea at the SCU allowed her to receive and benefit from the treatment. As a result, though she reported she "did not know what the future [held]", for now, she faced no challenges keeping up with her child's medication.

Another parent (Female, 40's) saw participation in genetics research as a way to receive more information about her child's condition. She explained that, when her child was born, she presented with characteristics that may have been indicative of a particular syndrome. She went on to state, "Basically, [they're saying] it cannot be proven or tested out here basically what type of syndrome she have."

She therefore believed that by allowing her child to participate in research, she would gain access to the resources needed to learn about the nature of this syndrome.

However, these sentiments emerged more commonly in reference to gene-based cures for sickle cell disease. Providers in particular explained that, as the cost associated with these treatments were likely to be "astronomical", Jamaican patients would not "be

able to afford [them] without some kind of affiliation with a trial center." Others maintained that most patients would only benefit from these cures "in the form of a clinical trial".

Education and Employment

From participants' discussions about the challenges and limitations SCD patients faced, issues related to education and employment emerged. Respondents explained that, as a result of the pain and leg ulceration associated with SCD, patients' education often suffered. One provider (Male, 30's) explained,

> "You find person's feeling pain they might not go to school today. And that can go on for an extended period where they miss school very often"

Providers went on to explain that the time spent "visiting to the clinic or to…the private doctors to get treatment" often resulted in repeated absences from school. Once again, these issues appeared to be have greater impact on those from rural parts of the island. One patient (Female, 40's) explained,

> "I grew up in the rural part of the country in Jamaican and going to school, I had to be in and out of school, missing school to go to Kingston for my appointment"

Some respondents also noted a phenomenon in which students tended to fall ill during examination periods. This would either prolong the amount of time it took for

them to complete their studies or prevent them from completing them all together. As one provider (Male, 30's) shared,

> "I've met quite a few, as I said we have the scholarship, who for their scholarship for their tertiary education, they don't take the average three years they take more than the three years to complete it because for some they get so sick that they have to go part time or do less courses because of the additional stress that it brings on or because they missed an exam or two so they have to come back the following year to do it and then again, they probably get sick again."

This was one parent's (Female, 40's) unfortunate reality. Her child, she lamented, always fell ill "whenever time {it was] to do some subject or some exam."

Respondents described the overall effect these challenges had on patients' education. One provider (Male, 30's) explained that, due to SCD symptoms, some patients were forced to "drop out of school", whereas others "finished high school without any qualifications." The latter was the case for one patient in particular. This patient (Female, 40's) explained,

> "…because of this, I wasn't able to do Common Entrance. And you know, Common Entrance then that would have taken you to a high school and then I went to grade nine and then all age school. From there, I did Grade Nine Achievement Test and 9 I wasn't successful either because of the sickness and not going, being in school."

Respondents recognized that limited education also often translated to poor employment prospects. “Due to their minimum education”, one Provider (Female, 40’s) explained, patients were often “not able to get a job that [could] sustain them.”

Disease complications, however, often impacted patients professionally in more direct ways. These complications prevented them from working or made it difficult for them to maintain a stable job. One patient (Female, 30’s) described how her experiences with leg ulcers resulted in unemployment. She stated,

> “When the leg ulcer starts on my foot, I used to stress it, like I would say ‘chu’, I won’t let this stop me from work. But when I saw it go two months, I was saying no man why is not getting better? Then 3 months, 4 months, 5 months, 6 months come I was saying man. When you see I had to stop the work that I doing because of this sickness I said Jah know.”

Others were unable to do the “kind of work that they’d like to do” as they required “a lot of stress” that they couldn’t “physically manage”. One parent (Female, 40’s) worried that SCD would limit her child in this way. She feared that, due to his illness, he would be unable to attain the job he hoped for when he became an adult. She explained,

> “I think certain kind of job he can't do. Yeah, because he always wants to be a pilot. I don't think he would, he could ever manage that, because there are time, there are time when, when you know it just come on him.”

Respondents saw unemployment as one of the main challenges that individuals with sickle cell disease faced. They went on to explain that this inability to find and

maintain gainful employment meant that many of these patients experienced poverty and financial restraints. Providers explained that many patients were unable to "to feed their families, take care of themselves", or support their children in attending school. Moreover, those who hoped to further their education were unable to do so due to these financial challenges. One patient (Female, 30's) who expressed her desire to "go back to school, [get] some subjects and help [her] son in the best way [she could]," confessed that her financial problems made achieving her goal difficult.

Financial Assistance

Respondents described sources of financial assistance that individuals with SCD could rely on. These sources supported them with both SCD treatment and management, and everyday expenses. When discussing the kind of financial support patients received, the PATH program emerged as one of the main sources. One provider (Female, 50's) explained,

> "You do have, for children, or for and adults, too, I think what's called the PATH program for people who…can't afford…who are of the lower, lower socio-economic background who don't, where they can, they can apply and get help, you know the children in school and, and so forth."

One Provider (Female, 40's) went on to say that this was a government program and though it provided some assistance, it was "limited as to what [patients] could receive." One parent (Female, 40's) revealed that with the program,

> "Sometimes you get…help, sometimes you don't because is not everywhere you can, the PATH, the receipt, is not everywhere you can use it. But it's good and, it's good sometimes to have it. And the next time you have to pay."

Respondents also described the National Health Fund as a source of financial assistance. Like the PATH program, this was a government program that "[reduced] a bit of the cost of some of their medication." However, this program also had its limitations. One Provider (Female, 40's) explained,

> "Unfortunately, as, as, the analgesics is not on that national health fund, and as I told you before, pain is one of the most common symptoms of sickle cell disease. And if analgesics are not on it, it is like almost no help at all."

One patient (Female, 20's) reaffirmed these issues. She reported, "They have the NHF but the NHF doesn't cover the medication that I want to buy. And sometimes I don't have the cash."

Finally, the SCU emerged as another source of financial support for patients with SCD. Respondents explained that the social worker often advocated for and launched fundraising efforts on the behalf of the unit's patients. The SCU also offered scholarships to tertiary level students that partially covered the cost of their education. One provider (Male, 30's) explained,

> "We have a scholarship, we have, it's funded by Flow. We have five persons per year, tertiary, post-secondary who gets up to $90,000 towards the tuition and it's been going on for the last um 10, 10 to 13 years."

Stigma

Stigma emerged in three main categories: enacted stigma, anticipated stigma and internalized stigma. These forms of stigma are defined in **Box 2.** Participants' experiences with stigma most often emerged from questions about the challenges patients and parents faced related to SCD and how respondents felt people with SCD were treated in their communities. Stigma was also associated with attitudes towards genetics research and cures for sickle cell disease.

Box 2: Types of Stigma

Perceived Stigma*:* Perception that one is being treated differently due to stigmatized status

Enacted Stigma: Experience of negative treatment or discrimination due to stigmatized stat**us**

Anticipated Stigma: The expectation or fear of negative treatment due to stigmatized status. Can manifest in anxiety about disclosure

Internalized Stigma: Adopting society's negative perceptions of stigmatized status

Participants' experiences with stigma are summarized in **Table 7.** When asked whether they felt that they or their children were treated differently due to their sickle cell disease status, 60% of patients responded affirmatively while all parents gave a negative response. Most respondent, however, reported instances of stigma in the community or interpersonal relationships, in the workplace, or in the healthcare system.

Table 7: Perceived and Enacted Stigma Among Patients and Parents

	Patient n=10	Parent n=9	Total n=19
Perceived Stigma			
Yes	6	0	6
No	4	9	13
Enacted Stigma			
Community & Interpersonal Relationships	10	5	15
Health Care	1	0	1
Work	3	0	3

Enacted Stigma in Community and Interpersonal Relationships

Respondents most often described enacted stigma that originated from the wider community. Much of this stigma came in the form of disparaging remarks, stereotypes or misconceptions that targeted individuals living with sickle cell disease. Respondents explained that patients were called names such as "sick foot", or "curry eyes". Sometimes, providers revealed, "they were called duppies" or "described as weak or sickly". Some were told that they were "not going to live past [a] certain age". Providers described a perception that those with sickle cell disease were "just worthless because they [didn't] want work" and that they would "never really amount to anything." Respondents further explained that stigma also resulted from fears of contracting the disease. As one Provider (Female, 40's) explained, this was particularly evident among school children who refused to associate with peers with SCD out of "fear that they [would] catch it from them."

Children with SCD were often ostracized by their peers. One patient (Female, 30's) shared that, while in school, she endured ridicule and ostracization by her classmates who were "afraid that [she] would drop down on them."

Some respondents, however, suggested that enacted stigma from the wider community was not quite so commonplace. One provider (Female, 50's) explained,

> "But it's not everybody, and I wouldn't say that, you know, because people have sickle cell disease that, we don't walk around with a sign on our head, you know people don't walk around with a sign on their head showing that they have it. So, from that aspect of normal everyday living, you may not be treated very differently."

Those without the outward symptoms of the disease were less likely to face this sort of stigma or discrimination. When asked whether his child was treated differently, for example, one parent (Male, 30's) replied, "no, because if I don't tell you that he has sickle cell, you won't know, right?"

Another parent (Female, 30's) echoed these sentiments. She explained that as her child didn't "look different" and she "[looked] normal" she had not experienced stigma. Similarly, some patients also explained that they did not "fit into that description" that others had of those with sickle cell disease, when asked about their experiences with stigma and discrimination. As such, they were not subjected to the mistreatment experienced by those who did. For some patients, this description included yellow eyes,

leg ulcers or frequent pain episodes. For some others, this description was simply "short or skinny".

Patients who experienced pain often experienced more stigma. Some of these patients reported that those around them would disregard or minimize their pain experiences. As one patient (Female, 30's) explained,

> "Sometimes when I'm feeling pain and people around me, they actually behave like I am pretending, like I'm not feeling pain."

Another (Female, 30's) stated that those around her blamed her for her illness. She stated,

> "And then I've been around persons where they would let me feel if I wasn't the person that I am they would actually let me feel as if it's my fault, as to why I'm sick or I have control over my illness and not doing anything about it."

Those with leg ulcers in particular faced a great deal of stigmatization. As one provider (Female, 50's) explained,

> "And then leg ulcers give another stigma, because you know people have this wrap up foot, sore foot, and people are like Lord them just is a sore foot sickle cell person because you know that, that, that is another thing."

These patients were teased, scorned, or became the subjects of unwanted attention. As one patient (Female, 50's) revealed in describing another patient,

> "I remember working somewhere and the lady, she was a sickler and had a sore on her foot, you know. So, you know, people always wondering, why you always have a sore, that sort of thing. "

A few adult patients also complained that they were often overlooked due to their small statures. One male patient (Male, 20's) explained,

> "When other people like enter events or certain buildings and they will find them as a threat, basically if you are a sickle cell patient and you look a certain way, even if you have weapons or something, they will not search."

Furthermore, they often felt disrespected, even by those younger than they. One patient (Female, 30's) shared,

> "When you go around some teenagers and when they look at you and you, like you said, you know that I'm older than you? And they say which part of you and those things (laughs) and…you have some of them big in body you know and just we're just little and like, I will say … I'm 28 and she will say, but I'm 16 so how you look so? Those little things. So, we have to sit, and we have to explain every minute. I don't like it."

To a lesser extent, respondents described enacted stigma in close interpersonal relationships. This most often emerged in the context of romantic relationships. Patients expressed that their partners often did not understand the challenges associated with and the implications of sickle cell disease. One patient (Female, 20's) explained,

> "All right, meaning admission in the hospital for a while. But other persons, it's like they don't understand, and like

> they can't wait, I'm taking up their time and. I said okay, go ahead."

As a result, this patient confessed, she was no longer "interested in [relationships] right now."

Another patient (Female, 30's) revealed that, due to her illness she, at times, would have little interest in talking to or in engaging in sexual activity with her partner. As a result, she went on to say,

> "My partner would be offended about it…I think he, I think don't understand what it mean, I think, at times, he think that um, probably I'm out there cheating, that's why I don't want to have sex with him or anything"

Others stated that their partners or potential partners had no interest in being with a person with an illness. One provider (Female, 20's) explained that some male patients struggled to find stable partners due to their issues with leg ulcers or priapism.

Finally, to a lesser degree, respondents described enacted stigma in family relationships. One provider (Female, 50's) recounted an experience she had had with a male patient who "had been emotionally and…physically abused by an uncle who made him feel very worthless and…told him he was not a man that, you know, he [would] never be a man."

Enacted Stigma in the Workplace

Individuals with SCD also experienced stigma in the workplace. Respondents indicated that patients often had difficulty acquiring employment due to employers' reluctance to hire those with the disease. One provider (Female, 30's) stated that recurrent illness caused employers to think of these individuals as liabilities. She explained,

> "They can be apprehensive when it comes to hiring someone with sickle cell because unfortunately if persons are recurrently ill or decide to miss work blaming their sickle cell or say that they can't do certain things because of their sickle cell…then that means that they're being paid depending on what type of contract that they have they're being paid to not do the work and the work still needs to get done. So, some employers are just would just err on the side of not hiring if they know that the person has sickle cell."

In other cases, another provider (Female, 60's) explained, patients were not given the position or "the promotion that they deserved" either because "they [had] been away from work too long, or…they [were] too sick", or due to doubts about their ability to safely and effectively perform the job.

One provider (Male, 30's) recalled instances in which patients were terminated from their positions when their employer realized that they had sickle cell disease. He explained,

> "I have persons who have said they have lost their job when they found out that they have sickle cell disease because the boss will not pay for your sick days, if you've being so sick

> so often. It's going to cost me more than just trying to find somebody who doesn't have sickle cell disease."

Patients also shared their experiences with stigma in the workplace. One patient (Female, 30's) shared that, while interviewing for a position, she disclosed to her employer that she had sickle cell disease. The employer, she explained, worried that the disease was contagious and had to be assured otherwise. She went on to say,

> "And then she has me there and I was working with her and then I go in crisis and then, I'm not quite remembering what she said but she did rough about it and treating me a way until I even started to cry and then after, she did let me go."

Providers also explained that mothers of children with SCD faced similar challenges. One (Male, 30's) stated, "People are afraid to hire parents who have kids with sickle cell, cause that they know they're going to be absent a lot."

Enacted Stigma in the Healthcare System

Providers reported that patients with sickle cell disease experienced stigma in the healthcare setting. Hospitals sometimes labelled these patients as drug seeking and as such, neglected their complaints of pain. Providers relayed accounts of this stigma. One provider (Female, 40's) stated,

> "I mean I have witnessed doctors and nurses offering placebo. Um, you know, sterile water for injection, rather than active ingredient analgesics for, for very obvious pain that's, that's going on. I've, I've witnessed nurses and doctors who have been unwilling to, to prescribe opioids for, for severe pain crises, or for chronic analgesic therapy

> at home because they're afraid of either addiction or, or overdose or, or whatever and patients that have organic need and, and whatever for the, these drugs"

Some providers allowed that some patients did become addicted to the pain medicines. But, as one provider (Female, 50's) explained,

> "Because the nature, the nature of the disease, is, is pain. Pain is a big part of it, and then the nature of some painkillers, is that they hook you. So, you know it's not, not the patient's fault, it's, you know what I mean?"

Nonetheless, providers went on to say, "a lot of the hospitals that [were] frequented [tended] to kind of lump all the sickle cell patients together" and so, those that were "genuinely in need of the pain medications" did not receive the required care.

However, patients or parents generally described their experiences in the healthcare setting as positive. They explained that, when they visited the hospital and revealed that they had sickle cell disease, they were seen and treated quickly. One patient (Female, 40's) revealed, "Whenever I go like to the hospital and once they realize that I am a sickler, I tell them, I will get through quickly."

Anticipated Stigma

Anticipated stigma also emerged from the respondents' description of their SCD experiences, and from their attitudes towards genetics research. Participants perceived health and illness as private matters. One parent (Male, 30's) drew on current experiences

with the COVID-19 pandemic to describe fears of disclosure through genetics research participation and introduce health-related anticipated stigma,

> "Maybe they don't want to be like, like how all Corona now, people stigmatize against someone when them hear say them have corona virus and like stay away from them. They don't want nobody…see them less of a person"

Most providers and some patients and parents described fears of disclosure. Patients worried that sharing their sickle cell disease status would result in mistreatment or discrimination. This was evident in cases related to the workplace and interpersonal relationships. Providers stated that patients often expressed reluctance to disclose their SCD status to their current or potential employers. One provider (Female, 60's) explained,

> "Persons who are usually, when they go for job interviews refuse to identify themselves as having sickle cell disease for fear of not getting the job."

To a greater extent, respondents described this fear in reference to romantic relationships. Some patients feared that if they told their partners that they had sickle cell disease, they would leave them. One provider (Female, 20's) spoke of a patient who had not disclosed to his wife "because he [believed] that if he [did], she [would] go."

Male and female patients expressed unique concerns about disclosure. One patient (Female, 40's) confessed that she feared telling her partner that she had sickle cell disease. She explained that, after giving birth to two children, she received a tubal litigation due

to her SCD status. She therefore feared that if her partner learned of her inability to have more children, he would leave her. She revealed,

> "…you know persons with sickle cell are not supposed to have children, much less two and I have 2 so the doctors tied my tubes and um after doing that, that has stopped me that as in the me from having, from being in a relationship that I would want to be in for a long-term relationship because, you know, sometimes you're in a relationship and the person realizes that you can't have any more children so they like, they're not staying or whatever. But, for this one, the person doesn't know that I took tie off. And the person doesn't know that I have sickle cell because I'm scared. I don't want to tell him, and then there he goes. He only has one child, and I only have two. So, I'm scared that he might want one more. And I'm scared that if I, if he found out that I have sickle cell then that's it so."

Male patients on the other hand, as one provider (Female, 20's) explained, worried that "their partners would [not] see [them] as a total man, a whole man."

For some, anticipated stigma resulted in self-limiting behaviors. Providers explained that school children with leg ulcers, for example, did "not want to participate in physical education classes" for fears of exposing their leg ulcers and being "ostracized or stigmatized or called names." Children also isolated themselves from their peers for fear of being teased or rejected. One provider (Male, 30's) explained,

> "Because the stigma that comes with it is a lot so you don't want to be ostracized or stigmatized or called names. So, they tend to, they stick to themselves."

In older patients, one provider (Female, 60's) explained, those with leg ulcers struggled to find partners because they feared that "persons may not want to talk with them [or didn't] understand about their disease"

Internalized Stigma

Most patients showed signs of internalized stigma. These emerged from discussions about their experiences with the disease, the ways in which they believed the disease limited them, and what they hoped to experience if they could be cured of SCD. Specifically, respondents often spoke of normalcy and normal people, usually in opposition to themselves. Many patients shared that they had "never really lived a normal life." One patient (Female, 40's), for example, explained that her pain crises "hindered [her] from being in school every day as a normal child would have done." Another patient (Male, 20's) explained,

> "In the past when normal people are doing exercises running or lifting up heavy objects, even though I can probably lift the same amount as them I can't do it as long as them."

While none explicitly described themselves as abnormal, respondents spoke of 'normal' as something beyond themselves, something they believed they were not. These patients compared themselves to those they believed were normal. These were individuals who appeared to not share their challenges or limitations and who had capabilities they aspired to. One patient (Male, 30's) confessed,

> "I felt like I wasn't normal like any other child and my, stuff that I really go through, normal people don't really go through stuff like that. Like most time I want to go to the beach and stuff like that and would have problem with the cold water and stuff like that. So, it was all it was. It was quite a challenge, growing up as a child, knowing that I have it."

These feelings of abnormality often resulted in depression among patients. One patient (Female, 30's) explained,

> "…like during my teenage years. I used to feel some form of like sadness or like depression, thinking that I'm seeing other persons like, they are normal and I'm not normal."

As previously described, patients spoke of a cure for sickle cell disease as the key to "a normal life". These patients hoped for a treatment where "[they wouldn't] have sickle cell anymore and [they would] be able to live a normal life."

As indicated previously, 4 patients, when asked if they were treated differently due to their sickle cell disease status, responded negatively. Notably, most of these patients, when probed further, recounted instances of experienced stigma or discrimination. However, it appeared that these patients had not internalized this stigma. While half of these patients mentioned 'normal' in relation to themselves and their experiences with sickle cell disease, these patients asserted that they lived normal lives and did not indicate that, at any point, they believed otherwise. Notably, also, three of

these patients revealed that they were not interested in receiving cures for sickle cell disease.

Infertility Stigma

Respondents also discussed the social consequences of infertility in women. Providers explained that many women struggled with the belief that sickle cell disease caused infertility. Many of these women were told that people with sickle cell could not have children. Those who did were often looked at as rarities. One patient (Female, 50's) who managed to become pregnant and have children revealed that her obstetrician was shocked to learn she had SCD. She explained,

> "He said how come you have sickle cell and I can't see it?…Him say but you don't look like you have sickle cell. Because look at you, a person with sickle cell and you so healthy and pregnant"

Another patient (Female, 40's) confessed that she saw herself as "a living testimony of sickle cell" because she was able to "have two beautiful teenagers."

In some cases, patients were told that they should not, rather than could not become pregnant. As one parent (Male, 30's) explained of his child's mother,

> "…the child's mother, a SS sickle cell she did have. So, doctors never did too want her to get pregnant. Them ever a say it risky."

In these cases, women who became pregnant sometimes faced mistreatment by providers who saw their pregnancies as reckless or irresponsible. One patient (Female,

30's) shared that a friend with sickle cell who had become pregnant experienced some complications while at the hospital. As a result, providers neglected and disparaged her for her choice to have a child. She explained,

> "Like when she's crying and bawling for pain, they don't pay her any mind. And like she would just be, the same thing as what I said like they would say "you gone breed and you mustn't breed"

Many women who became pregnant, however, suffered miscarriages. Some, as one provider (Female, 20's) stated, experienced as many as 4. One patient (Female, 30's) shared her experiences with miscarriages. She explained,

> "Each time I get pregnant, I have a miscarriage. I don't know what's the cause for it… I tried, what, four times and I had four miscarriages."

Some women became pregnant despite the significant risks it posed to their own health. One provider spoke of a young woman with a complicated form of sickle cell disease who was adamant about having a child with her partner. The provider explained that she tried to convince her to delay her pregnancy so that she could safely take hydroxyurea in the hopes of improving her health. However, this was to no avail. She went on to say,

> "She eventually did become pregnant…and unfortunately, the worst that I had predicted happened with her and she ended up in the ICU and in multi organ failure and, and she ended up dying in the ICU."

Women who struggled with infertility faced significant psychosocial consequences. They faced stigma within their communities and in their romantic relationships. As described above, women with sickle cell disease experienced anticipated stigma, fearing that disclosure of their status and their inability to have children would cause their partners to leave them.

Respondents also detailed instances of enacted stigma due to infertility. Providers shared that these women "would tell you that they [were] looked down on in their communities, because they [couldn't] have children or if they [lost] a pregnancy". One provider (Female 20's) described one patient in particular "who said that her partner was about to leave because she wasn't able to have a child." One patient (Female, 30's) shared that a previous partner tried and failed "for about five to six years…to get [her] pregnant". She explained, "he used to ask me if I'm a mule or I'm a barrel or stuff like that."

Women who wanted to have children but were unable to, faced a high risk of depression. Providers explained that these women sometimes "broke down". Some patients conveyed a sense of despair. One patient (Female, 30's) in particular, revealed that she had given up on trying to get pregnant. Later, when asked how she imagined her future she confessed that she wished it were brighter. She then went on to say, "Hopefully to have a child sometime. Hopefully if it's possible. By the help of God"

The ability to have a child was highly valued among women with sickle cell disease. It appeared to be linked to the value of the woman herself. Notably, however, this social expectation appeared to contradict the emphasis respondents also placed on decreasing the incidence of children born with sickle cell disease. Providers explained that they encouraged patients to bring in potential partners for screening, with one provider (Female, 60's) stating,

> "Because at the end of the day what we're trying to do is to, to educate and be hopeful that the incidence of sickle cell disease can be reduced through education."

One patient (Female, 40's) who had had two successful pregnancies at times seemed in conflict with her choices. She asked that individuals with sickle cell disease,

> "Be careful. Unlike me, I have two teenagers. Both of them have the traits. So, what I do. I told them whenever you're having a partner, make sure you get your partner tested before you have kids"

She went on to say that patients should,

> "…make sure they do the genetic thing, then it would be easier because they will be able to know that all right, I am not willing to be involved with this person, because I don't want my child to come and have this thing [that]…would hinder her from growing into a normal child."

Another patient (Female, 30's) stated that she coped with her inability to have a child by looking "at it like the positive side". She explained,

> "…look at it as suppose the child would come with sickle cell disease and sick, sick like me. I could not bear that. Couldn't manage that."

While these women wanted to have children, they wanted to ensure that their children were not born with the disease. They however recognized the relatively high likelihood that this would occur if they did not ensure that their partners did not have the trait. Moreover, some parents, as one provider (Female, 50's) explained, experienced guilt over having a child with sickle cell disease. She stated, "sometimes some parents have a feeling of guilt, like they felt that they gave it to their children."

Cultural Issues

Finally, a number of cultural issues emerged from participants perspectives on genetics research and cures for sickle cell disease, and their descriptions of their overall experiences with SCD. These issues included influence and information, faith and spirituality, and attitudes towards health.

Influence and Information

Patients and parents of patients obtained sickle cell disease information from a number of sources. This included information about SCD cures and SCD management in general. These sources also influenced patient and parent decision-making. The most significant of these sources was the SCU. Providers alluded to the relatively paternalistic nature of medicine in Jamaica and described patients as heavily dependent on medical

professionals for information and guidance about treating and managing their disease. One provider (Female, 30's) explained of her patients,

> "…the health care provider is, also a very, they look to us a lot for their decisions to be made, they'd like, you know, Doc, what do you recommend which one you think is best for me? Do you think I should do this?"

One provider (Male, 30's) described this as the "yes doctor syndrome." He explained, "If the doctor says x, they're going to go with what the doctor says."

Providers also explained that even patients who had gone abroad would contact them to ask their opinions on the care they were receiving from their providers overseas. One provider (Female, 60's) recounted,

> "I have patients who will call me from overseas, just to find out from me you know, if the doctors over there are doing the right thing, because they have a confidence in the staff here at the sickle cell unit. Even today, I had a patient calling me to ask my opinion on bone marrow transplant."

Patients and parents often proved this to be true. When asked where they obtained information about sickle cell disease, all patients and parents reported that the SCU was one of their main sources of information, and therefore influence in SCD disease management. One patient (Male, 30's) expressed, "You still would have to depend on a doctor to tell you what type of drug to take and stuff like that in order for you to get healed."

Indeed, for many patients and parents, the SCU was their first source of information about sickle cell disease. Many confessed that they had known very little about the disease and the challenges that might come with it, prior to their first visit to the unit. One patient (Female, 30's) revealed,

> "Actually, I did not have any information about it until I joined the clinic by UWI. I used to go to doctor and only know that I have sickle cell. I never know what it actually means when they said you have sickle cell. What is cause, only just know that I have sickle cell."

Upon his first arrival at the clinic, one parent (Male, 30's) explained,

> "They gave us some brochures to read and them things there. The doctors them always a tell you, you know, every time you go. Them always a give you some advice. The doctors them up there and the nurses."

Providers theorized that patients' reliance on the SCU for information and advice stemmed from the trust that providers cultivated with them. As one provider (Female, 60's) explained, "Way back in the 50s, 60s…very little was known about the disease." However, when the SCU became the specialist clinic in Jamaica, patients felt "they had a place for themselves, where they were understood." This provider went on to reason that this sense of being understood was instrumental in patients' and parents' trust in the SCU. Patients believed that providers were working on their behalf and looking out for them. The nature of this trust, respondents explained, was such that patients would seek

guidance from the SCU even for issues not relating to sickle cell disease. As one provider (Male, 30's) revealed,

> "If something happens, let's say, something that is not sickle cell related they're going to come to the doctors at the sickle cell clinic to talk to them about that condition instead of going to let's say a GP or something. So, they think that they're the ones to run to for anything that they have."

Patients affirmed these claims. One patient (Female, 50's) explained,

> "If ever I feel sick and I just suspect it might be the sickle cell, even if is not the sickle cell, I always check them. I just have confidence in them (laughs) I have confidence in my general doctor too but I just always like a second opinion seeing that they know my history with the sickle cell."

Providers often developed close relationships with patients and parents. They confessed that their relationships sometimes blurred the line between professional and personal. One provider (Female, 40's) who worked on the unit's research side described the SCU and their patients as "like a family". She went on to say that the clinic gave these patients "a sense of belonging", and people they could rely on for information and assistance.

Providers however often emphasized to patients the importance of obtaining information from other reliable sources. One provider (Female, 60's) explained,

> "We advise them to, to make sure that you go to reputable sources to get their information. In other words, grandma isn't a doctor grandma isn't a nurse. The man down the road is not a medical person. So, we make sure to point those

things out for them, when their seeking information to go to the right source."

Providers posited that information seeking habits were tied to an individual's education level. One provider (Female, 50's) explained that, while most of their patients or parents deferred to the guidance of their providers at the SCU, "some [would] do their own little research depending on educational level". This was evident among this study population. Patients and parents with tertiary level education tended to report using more information sources than those with secondary or primary levels. Moreover, more patients and parents with tertiary level education named the internet as one of these sources compared to those with only secondary or primary levels (**Table 8**).

Table 8: Comparing SCD information sources used by patients and parents by highest education level

	Primary n=1	**Secondary n=11**	**Tertiary n=7**	**Total n=19**
Friends &Family	0	3	2	5
Internet	0	5	5	10
SCD Peers	0	4	5	9
SCU	1	11	7	18

These parents and patients described using the internet to obtain general knowledge about sickle cell disease. They used websites such as Facebook and YouTube and carried out general searches on Google. One parent (Female, 30's) explained that she

often engaged with SCD stories and articles recommended through these platforms' algorithms. She revealed,

> "What I see about the internet is if you follow one topic, you always see pop up, pop ups come in, wherever you go on Facebook on Google, the thing, you see the same things popping up topics about the same popping up popping up so eventually I just read them, and they just keep coming in."

Others turned to these sources to obtain specific information about certain topics. At times, they sought information about treatments their doctors provided. As one patient (Female, 30's) shared,

> "Suppose I take this medication and I start to feel weak or something and on, when I start to read up on a medication, it doesn't say weakness, I will stop taking it. And then I'll go back to the doctor and tell the doctor and explain to the doctor. And they might even give me something else."

Another parent (Female, 30's) revealed,

> "I also like, Google, go on YouTube and, and watch videos and stuff like that. Cause if he, if he started because, when he was starting the HU, even though they gave us, they gave us um, stuff to read and stuff like that, I, I went and did my own research."

At other times, they sought information about new or potential treatments, or cures for sickle cell disease. One patient (Male, 20's) explained that, while his providers at the SCU "[told him] all [he needed] to know, and the drugs that [were] available", he

believed they held back information about experimental drugs or treatments. He would therefore turn to resources like YouTube to access this kind of information.

These platforms provided a means for these individuals to play a more active role in their or their children's health. However, as one provider expressed, these resources could sometimes prove dangerous, "depending on how well-equipped people [were] to be able to research where information [was] coming from [and] where it [had] been published."

They worried that some of these sources offered patients misinformation and disinformation that could be harmful and act as barriers to care. Conspiracy theories that spread through these platforms bred mistrust and, for example, led to reluctance towards certain essential medications or to taking part in genetics research, as previously described. One provider (Female, 40's) explained,

> "Because there are a lot of like patient groups on you know on Facebook and Twitter and stuff like that and, and I, I am on some of them. So, I can see what, what people are writing to each other. But there's, there's a lot of conspiracy theories out there that, that fly around."

Providers therefore emphasized the importance of patients and parents knowing how to "wade through this information."

Concerns about information quality were not limited to online sources. Patients and parents also turned to family and friends for information and advice about managing sickle cell disease. Many patients explained that their parents were instrumental in their

SCD education. One patient (Male, 20's) revealed that his mother often relayed to him SCD information that she had found or researched. When asked how she first came to understand sickle cell disease, another patient (Female, 20's) simply stated, "well, my parents educated me."

Others sought information or advice from other family members. For example, one patient (Female, 20's) explained that her sister had told her about bone marrow transplantation and another (Male, 30's) shared that his sister sometimes gave him advice on how to manage the disease. One parent (Female, 30's) revealed that she tended to look to her uncle for information about treatments her providers prescribed for her children and to "elders in [her] family" for knowledge on traditional medicines.

Notably, information from family often emerged in the context of traditional or alternative treatments. As one patient (Female, 30's) explained, her mother pushed her "to eat the beet root, to eat the callaloo, to eat this to eat that" in order to treat certain disease symptoms. Providers reaffirmed this phenomenon, while also expressing concerns about the information's quality. One provider (Female, 60's) stated,

> "Because I do find, for example, you may have a grandma in the country, who will tell you, and I'm speaking in particular to leg ulceration…you find, many times that they go home and we advise the patients to do you know to do this particular treatment or so on and they go home and there's a grandma in the country who tell them no. They must not do that. They must use this bush or they, you know, we do get an interference. And they will listen."

While children naturally relied on their parents to make decisions regarding their health, adult patients also tended to depend on parents and other relatives to provide information and guide them in disease management. One provider (Female, 40's) explained,

> "Our patients because it is a genetic disorder you have from birth, right, so the parents are the first and foremost, parents, parents are the first and foremost that make the decisions for the child. So, the child grows up knowing that the parents and family members make decision, help in the decision-making process. And as they get older, then they will tend to rely, still continue to rely on that."

As a result, providers found that patients sometimes returned after consulting with family about a particular treatment course to "say this person said no."

Some patients, however, emphasized their need to screen the information and advice they received. One patient (Male, 20's) explained that,

> "If you can't experience it like I how do, and so if you don't have the experience already then basically to me your information is not really that helpful."

Instead, these patients either relied on their own insight or sought information from their peers whom they believed could relate to their particular challenges. One patient (Female, 30's) stated that she sometimes turned to "other sicklers" for advice because "they [were] sicklers like [her] so [she liked] to get their perspective on the

illness." Other patients looked to them for information and advice on medications. One (Male, 20's) patient explained,

> "I talked to a friend in England, and they suggested different medication, what they had taken growing up for sickle cell and they sent it to me."

For parents of children with sickle cell disease, other parents in similar positions were instrumental sources of information, advice and influence. This was particularly so in relation to the child's medication and treatment. Providers explained that parents of children prescribed hydroxyurea, for example, often displayed reluctance or skepticism about the drug, due to the listed side effects. However, one provider (Female, 30's) explained,

> "I've seen where other parents who have children on it would speak to them and say, no, you know, try it, you know they'll, they'll, they have, they have done so much better since they've been on it."

These parents, one provider (Female, 20's) went on to explain, influenced by "another parent's testimony", often returned to say, "okay, I want it for my child."

One patient (Female, 30's) shared a similar story. She revealed that, upon first hearing of bone marrow transplantation, she was concerned about the safety of the procedure. However, she explained

> "When I heard the other persons, not even with sickle cell, but other patients then that would have done it is okay or they are okay. I remember going to a seminar once and the

> first fish always sick so that did it, and she was okay, and I heard her speech. So, then I kind of developed, like have a more open mind towards it than actually before."

Nonetheless, respondents reported that patients and parents tended to take the information they gathered from outside sources back to the SCU for appraisal. One provider (Female, 30's) explained,

> "Some of the patients actually come to us and will say somebody told them about this treatment, or they were reading online and they saw this option and they will come back and ask us what our thoughts are on whichever treatment option they have found."

One patient (Male, 30's) confirmed this, saying,

> "I usually run it through them first to see because they're the specialists and I want to know what say. And basically, that's the main way I could get um factual information"

For some parents, children played an instrumental role in SCD related decision making. Children that asserted some degree of independence over their health tended to have some input in their disease management and treatment. Providers explained that they encouraged patients "to be a part of the management and the decision making" during their teenage years.

One parent (Female, 40's) spoke of her 15-year-old son, who exerted some degree of control over his health. She stated that he had "a full understanding of what he must do or not do" in order to manage his health. She went on to say that he had "been reading

up a lot on sickle cell and the cure". When asked about deciding whether or not her son would participate in genetics research, she explained,

> "If he is interested in it, he will do, he will do whatever he's told if he's interested. But if he doesn't want to do it, he's not gonna provide anything but if he if he's interested in it, he will do it."

Another parent (Male, 30's), though his child was much younger, shared similar sentiments. His son, he explained, also displayed independence in his healthcare and disease management. He monitored his medication and ensured that his supply was maintained. When asked about the kinds of samples he would allow his son to provide for genetics research, he replied,

> "I would a just ask him which one him comfortable with. Cause even the other when, the last time I went to do um the blood test for him, a him a show the phlebotomist say no man you not going any no vein around there so man, see one nice one round here so. And she have to smile, she have to laugh like she a say she never see no little youth come in and a behave so."

Providers stated that independence sometimes translated to problems with treatment adherence in adolescent patients. This was particularly the case for hydroxyurea. Some adolescents who began the treatment as children, they explained, stopped taking the medication when they become teenagers. One provider (Female, 60's) hypothesized,

> "I think this is a time when majority of them are feeling well. They want to be like their peers, you know, they want to be, they don't want to be different. They don't want to have to be swallowing tablets everybody. They want to be free; they want you to do what others are doing."

Another provider saw this as adolescents (Female, 30's) "realizing that they [were] kind of in control or [wanted] to exert their control over their body" and explained that "whatever [wanted] to be discussed with the parents, if the teenager [wasn't] on board, then you [were] going to have a battle with regards to adherence."

Faith and Spirituality

Faith and spirituality emerged as an integral factor in sickle cell disease management and decision-making. Respondents described faith and spirituality as a tool often used to cope with the challenges of the disease. Providers underscored the importance of patients' faith in God in coping emotionally. Patients and parents found strength through their faith and used it to maintain a positive outlook. As one provider (Female, 50's) explained, "People pray, you know, and um, and I'm a believer in prayer, I believe that prayer helps to give you a little bit more strength."

Providers stated that patients gained power through knowing that "God [was] always on their side", that "somebody [was] helping [them] to bear [their] cross."

Patients reflected on the effect that faith and prayer had on their ability to "look at…the positive side", and to not be overcome by the challenges they faced. When asked

about the ability to remain positive, one patient (Female, 30's) replied, "God. Trust me, God (laughs) No one else but God."

Patients and parents also maintained that their faith was as much a source of physical strength as emotional resilience. Parents in particular believed that God protected their children and kept them alive in spite of their disease. When speaking of the role her spirituality played in her child's SCD management, one parent (Female, 30's) explained,

> "Even now, yeah, I think prayer works. Trust me. I don't think it works; I know it works. Cause sometimes some things that you go through and you see some people go through it, they don't make it out of it then you make it out of it, not by chance, I think, I don't think it's by chance."

For one parent (Female, 40's) , her faith in God surpassed her faith in conventional medicines. This parent affirmed that it was this faith, in combination with traditional treatments, that had kept her son alive,

> "Because without God in the midst I know he would have died long time. So, my faith in this, in God and in the herbal is more stronger than my faith in medical drugs or what would you say? Yeah."

One patient (Female, 50's) spoke more specifically about prayer's impact on her physical health. She explained that she had experienced knee pain for weeks and it was only after she prayed for relief that her pain dissipated and had not returned. She recalled,

> "So, one night, I really got fed up say Lord, no. I'm your child you have to do something about this. And I took the olive oil and I anointed it and I prayed, and my dear lady would you believe it or not, from that time this knee hasn't pained me. That is like probably 2, 3 years now. Yeah. No severe pain."

While faith often aided in coping, internalized stigma related to sickle cell disease sometimes manifested in patients' attitudes towards faith and spirituality. One provider (Female, 20's) and one parent (Female, 40's) described some patients' belief that their disease was a sign that God had rejected them. The provider explained, "Because some of them will tell you why would a good God do something so wicked to them?" The parent confessed that she had to "teach [her son] to pray" and to let him know that "even though he [was] going through the pain, it [didn't] mean that God [rejected] him".

For a few respondents, faith and spirituality colored perspectives towards SCD research and management. Though one provider (Female, 30's) maintained that belief systems did not play as great a role in disease management and decision making, another (Female, 50's) stated that in her experience, the opposite was true. She explained that those patients who were "church-going" or religious, tended to seek the perspectives of their fellow congregants. She claimed, people's religions were also likely to influence their attitudes towards genetics research and gene-based cures for SCD. This provider stated,

> "So…I think that would be, I think people's religion also. That's the truth. Because some people feel that oh, you mustn't interfere with anything"

Patients reaffirmed these perspectives. In describing others' reluctance towards genetics research and gene-based cures, one patient (Male, 20's) explained that some individuals were "conscious like that" and were afraid to do anything to "affect or change anything about them". This patient went on to explain,

> "…some of them will just believe that if they probably continue on the spiritual route then maybe one day, they'll get better."

Another patient (Female, 50's) added to this idea, this time, speaking of her own fears and hesitations about gene manipulation. She confessed,

> "I believe that god created us unique, you understand? And the genes is one of the unique, uniqueness about us."

She therefore worried that editing or otherwise manipulating a person's genes could have negative consequences.

A few parents, however admitted that, though they believed their faith in God had the power to heal, individuals still needed to take an active role in their health and take advantage of the resources available to them. One parent (Female, 30's) explained,

> "… 'cause God say him look out for people that look out for themselves. So, you still have to look out for yourself. You cant' just sit down and pray and say God a go have you. You still have to look out for yourself. Try stuff."

Attitudes Towards Health

Finally, respondents discussed their perception of and attitudes towards health. Providers, and patients and parents described health in fundamentally different ways. Providers described health as multifactorial, encompassing physical, mental, emotional and spiritual factors. Good health was therefore a state of wellbeing across all four dimensions. They approached the concept more clinically, and often cited definitions learned during their medical training. When asked to describe good health, one provider (Female, 40's) offered,

> "By WHO it's a state of emotional, physical and spiritual wellbeing. Yes, but I do. I mean, I do believe that but good health is multifactorial. It's not just physical health that we that we're talking about. I think it's, it's much more all-encompassing."

Most providers weighed these elements of health equally. However, a few viewed mental and emotional wellbeing as most important. For one provider (Male, 30's), these elements were the basis of health and necessary for wellbeing in all other facets. This provider explained,

> "Personally, um good health starts with peace of mind…You have to think the mind will always be with you. When you go to bed at night, you will be left with your thoughts. So, once it your mental state is not comfortable, you can't be comfortable."

One patient (Female, 30's) also viewed health as multifaceted. Like the providers above, this patient explained that health was not simply the absence of physical illness, but also the presence of physical and mental health:

> "Good health is also being mentally healthy, physically healthy, not just the fact that, okay, you don't have an illness, so you're considered healthy, basically. Good health is just being able to know that you're okay, generally. Mentally, physically, stuff like those."

For most patients and parents, however, perceptions of health seemed related to their personal experiences with disease and illness. They focused heavily on the physical aspect of health. Patients described it as being "in good condition", "physically fit", and "feeling good in your body." One parent (Female, 40's) associated good health with living "a longer, better life."

Patients and parents also had an emotional response to the idea of good health. They explained that good heath meant "the world", it meant "everything" to them. They associated good health with a life free of limitations, with being able to do everything they wanted or hoped to do. This was particularly evident among patients who felt limited by SCD and felt challenged physically, socially or emotionally by the disease. As one patient (Female, 40's) explained,

> "Good health means the world to me. Having good health is the best thing because I mean when you're not in good health, you're not, you're not able to function the way you would want to. So, having good heath means the world to me."

These attitudes were also evident among patients who faced fewer SCD related difficulties and recognized the instrumental role that good health played in their situations. One such patient (Male, 20's) who expressed that he had no need for a SCD cure stated of good health, "Good health? Like what I'm doing now, you know like living a normal life, um, healthy I suppose."

Patients and parents placed significant value on good health. As the previous quote demonstrates, they associated good health with normalcy and 'a normal life' or 'better life'. Ill health, therefore, was likely associated with an abnormal life or a worse life.

One patient (Female, 30's), however, emphasized that good health did not necessarily control one's outcome. A person believed to have good health could have worse outcomes than those believed not to. This patient explained,

> "Because true with how persons who would consider healthy stuff will happen to them and then someone who won't be considered healthy like someone with sickle cell, we are okay. So, um it's just basically seeing what happens on a day-to-day basis."

Providers, and patients and parents also differed on how they determined whether an individual possessed good health. Patient and parents tended to view good health as binary, the absence or presence of severe disease or illness. As one parent (Male, 30's)

explained, good health was, “When you not sick. Once you not sick (laughs). As in a clean bill of health, everything a work all right.”

According to one patient (Female, 50’s), good health meant “just being generally free of pain. Not having any chronic ailments.” Those who lived with a severe chronic ailment or who showed signs of physical disorder, therefore, did not have good health.

Providers, on the other hand, had a more nuanced view. They generally considered good health as a spectrum and relative to the individual. As one provider (Female, 40’s) explained,

> “It can vary, what we will consider optimal physical, mental and social wellbeing, what, what might be optimal for one person may not be for another.”

They maintained that control and trajectory defined good health. Those that were able to control the physical, mental and emotional aspects of their health to the point of being functional, or those who saw improvement in these aspects overtime enjoyed good health. They argued, therefore, that even those with serious chronic illnesses could, indeed, be in good health. As one provider (Female, 60’s) asserted,

> “Persons who have um certain um, some persons who are you know, have certain abnormalities, they can have a good health. You know, so it doesn't, it doesn't mean that you have to, it doesn't mean that you have to be all of everything to have good health.”

An ‘abnormality’ in one’s condition, therefore, did not necessarily equate to ill health or an abnormal life.

Notably, most respondents believed that anyone could achieve good health. While most parents and patients held this stance, providers were more divided on the issue (**Table 9**). Though patients and parents saw health as binary, they generally believed that individuals had control over which category they belonged to. Most believed that factors they considered modifiable like mindset and lifestyle were most instrumental in one’s health. As one patient (Female, 40’s) put it, “It’s all up to you. How you take care of yourself. How you exercise, what you eat, how you eat, when you eat. Your, your, your intake.”

Table 9: Participants' Perceptions of Good Health, by participant type

	Provider n=10	**Patient n=10**	**Parent n=9**	**Total n=29**
Is good health universally achievable?				
No	6	2	2	10
Yes	4	6	5	15
What factors influence health?				
Biology	7	4	1	12
Lifestyle	4	9	7	20
Mindset	5	2	0	7
Physical Environment	2	1	0	3
Social Environment	5	1	1	7

Patients and parents most often viewed diet and exercise as the main determinants of health. One parent (Male, 30’s) referenced his mother to explain this. He revealed that,

though she had sickle cell disease, she remained healthy. This parent attributed this to her diet. He stated,

> "She don't look her age and them thing there, so and that has to do with things she eats. Cause she know what to eat. She love her vegetables and them things. If it wasn't for her lifestyle, she wouldn't live so long. And she did have sickle cell."

A few patients viewed mindset as equally important. One such patient (Male, 20's) attributed his lessened SCD severity to his decision to not be limited by the disease. The patient explained,

> "Maybe it's because I participated in different things, me never just lay down and take it. It was just my drive. So, I don't know, for some people, it's a mindset."

A few providers also believed that good health was universally achievable. These providers also cited lifestyle and mindset as integral to good health. One provider (Female, 20's), when asked how individuals could achieve good health, highlighted the importance of mindset. This provider explained that one could achieve good health by "trying to stay positive. Positive no matter how, deep hard it might seem just stay positive"

Most providers, however, who generally viewed good health as a continuum, held that health was, for many uncontrollable. They allowed that while most people could achieve good health, some could not or would not reach that point of being functional, or of seeing steady improvements in their health. These providers believed that factors that

were harder to modify like biology, or the social and physical environment had the largest impact on one's health. As one provider (Female, 40's) explained, while lifestyle and mindset could influence health outcome for most, for those with significant biological concerns, this was not the case. This provider stated,

> "We may have control, especially when we don't have any chronic illnesses… if you don't have psychosis, or something that's genetic again, then you have control over that to, to have positive way, ways of destressing and eating healthy, having a well-balanced life."

Another provider (Female 50's) viewed good health as transient. This provider reasoned that while some may be able to achieve good for a time, they would not be able to maintain it forever. Good health was therefore, in some sense arbitrary and beyond the individual's control.

> "Not forever. It's relative to a time. I can, I can have good health today. I don't know what going happen tomorrow. But you know I can. And in terms of maybe my cardiovascular, but then I could develop a tumor somewhere else. So, you just don't know it's, it's in God's hands, I think, that, you know."

4. Discussion

Genetics research offers insights into SCD complexity, and technologies such gene editing and gene therapy are promising, new approaches to SCD management and treatment. Genetics research and these curative technologies, however, have important ethical, social and cultural implications and contexts that researchers and clinicians must consider. While H3Africa and other research groups are beginning to examine these implications in some African countries, to our knowledge, no studies have explored these issues in the Jamaican context. This study, therefore, explored Jamaican SCD stakeholder perspectives on genetics research and cures for SCD, and the social, ethical and cultural issues that relate to these perspectives. Understanding these issues, and using them to develop a region-specific framework, is an integral step to introducing these technologies to the Jamaican SCD population.

Participants' attitudes towards genetics research and cures for sickle cell disease, and the ethical, social and cultural factors that help to contextualize these attitudes are summarized in **Figure 4.** We propose this framework as a guide for conducting genetics research and implementing gene editing and gene therapy technologies to Jamaica's SCD population. This framework is explained below.

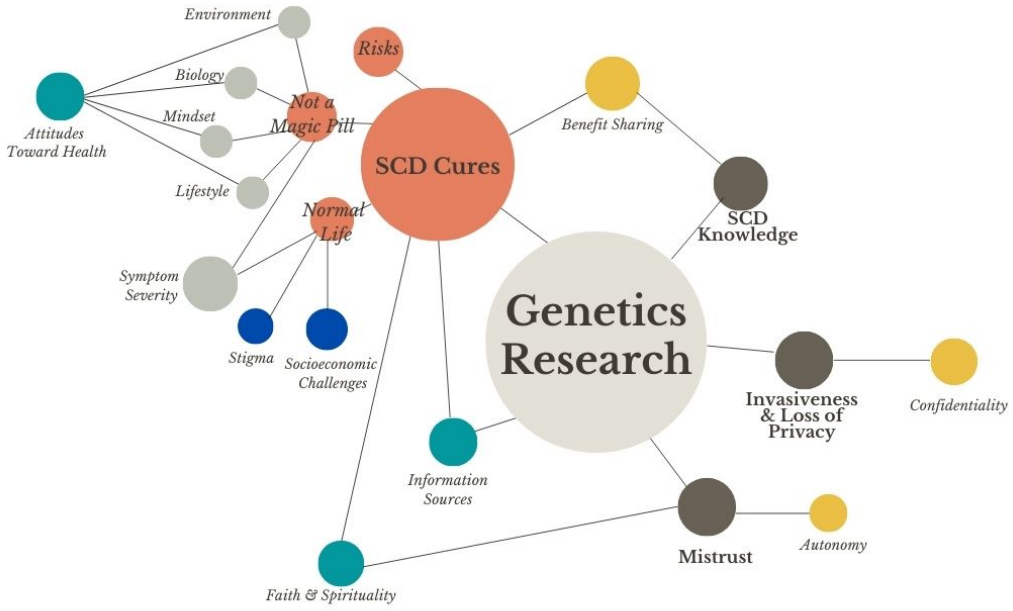

Figure 4: Proposed Ethical, Social and Cultural Framework

Participants' views towards genetics research are at the center of this framework. Overall, respondents viewed genetics research positively. They expressed enthusiasm and optimism about the promise of genetics research. They viewed genetics research as a way to improve both SCD treatment and management, and to advance SCD knowledge.

All participant groups hoped genetics research would improve the overall SCD knowledge base. Providers, in particular, emphasized the importance of Jamaicans' participation in such research in achieving this goal. However, many patients' and parents' interest in genetics research was driven by the personal SCD knowledge they believed it would provide. Patients and parents felt that genetics research would provide

a means by which certain health concerns could be diagnosed or better defined, and would offer insights into overall health and heredity. They also believed it would empower SCD patients to make informed decisions about partner selection and reproduction, and to ensure that their children and grandchildren did not face the same challenges they had. These attitudes towards SCD knowledge were similar to those seen within other communities. Previous studies found that Kenyan SCD stakeholders and Black African immigrant community leaders spoke of the benefits of acquiring personal SCD knowledge through research[22,23]. Likewise, they believed this knowledge would empower members of the SCD community to make informed health decisions and be better able to "manage future reproductive risks"[22,23]. Patients' and parents' belief that research participation would yield personalized results about heredity and individual health pointed to what Tindana and de Vries [24] termed diagnostic misconception. They defined this as a phenomenon in which participants conflated research with health screening or diagnostic efforts[24]. Relatedly, therapeutic misconceptions also emerged among some patients and parents where they seemed to conflate research and healthcare.

Providers, patients and parents also believed that genetics research participation would result in improved SCD treatment for SCD patients. Many participants understood that genetics research could yield a greater understanding of SCD and thereby result in the development of better options for SCD treatment and management, such as SCD cures. In this regard, many patients also expressed altruistic motivations for genetics

research participation. They hoped their participation in genetics research would create tools for improved SCD management not only for themselves, but for the wider SCD population. However, some patients and parents conflated SCD research with receiving SCD treatment. This became increasingly evident as many patients and parents discussed genetics research participation as if speaking about receiving SCD cures. They appeared to view these two concepts as one as the same, rather than seeing genetics research as a path to improved SCD treatment options. Many patients and parents therefore framed patient participation in genetics research around this therapeutic misconception.

Though evident in many settings, both therapeutic and diagnostic misconception have been widely described in relation to studies conducted in low- and middle-income countries (LMICs)[24-27]. Tindana, et al. [25], for example, described this phenomenon in participant understandings of the MalriaGEN study and their motivations for study participation. The presence of these misconceptions among certain patients and parents in the study, suggested limited understanding of the aims and methods involved in genetics research, similar to that observed in this present investigation. Therapeutic and diagnostic misconception could have significant implications for participant autonomy. They can create false inducement among study participants and result in confusion or unmet expectations[27]. Furthermore, participants who carry these misconceptions may not understand the implications of their consent. Particularly, confusion may arise regarding

key elements of genetics research studies such as biospecimen storage and future sample use[24].

Despite this generally positive attitude towards genetics research, respondents also expressed reservations about genetics research participation. Mistrust, and invasiveness and loss of privacy were significant points of concern among providers, patients and parents. Mistrust was most often described by healthcare providers. Providers, patients and parents described both their own concerns about misuse and mistrust, and concerns heard within their communities. Mistrust and fears of misuse often stemmed from misinformation about researcher motivations and spread by way of online sources, or throughout communities by word of mouth. Some participants, however, cited a history of abuse and misconduct in research that disproportionately affected Black peoples globally. Many of these fears were also anchored to current events. Parents, patients and providers used the COVID-19 pandemic and fears that the developing vaccines could harm rather than help, for example, as points of reference.

Participants also expressed concerns about invasiveness and loss of privacy through genetics research participation. Patients and parents in particular prioritized privacy in matters of family and health. They worried that genetics research could expose sensitive information about one's illness and family that they hoped to keep private. Parents, patients and providers discussed the social and financial ramifications of privacy loss for patients and their families. These included increased insurance premiums and

stigma in the workplace or in interpersonal relationships. Previous studies cited a similar urge towards privacy as protection from stigma, oppression or harm[22,28].

The proposed framework connects participants attitudes towards genetics research and those related to SCD cures (**Figure 4)**. As previously described, much of participants' enthusiasm towards genetics research was associated with their hope for improved SCD treatment options or SCD cures. All participants showed great enthusiasm towards cures for SCD. All providers and parents supported patients receiving SCD cures. However, only 60% of the patients reported interest in receiving a SCD cure. Although most patients had heard about cures for SCD in some capacity, patients and parents had limited knowledge about these cures. Moreover, most of these respondents had only heard of BMT. Patients and parents also expressed several misconceptions about SCD cures. Some worried that BMT required bone marrow from a dead body or that it had a success rate of only 50%. Other patients and parents shared concerns that gene-based cures would cause the recipients to no longer be related to their parents or children. Patients' and parents' knowledge and understanding of SCD cures was largely related to their information seeking habits. Those that had prior knowledge of SCD cures reported receiving SCD information from a wider range of sources including the internet, family and friends, and SCD peers.

Patients' and parents' attitudes towards SCD cures were largely related to the belief that cures would lead to a normal life for patients, the recognition that a cure was

not "a magic pill", and the perceived risk associated with them. Most participants believed that SCD cures would allow SCD patients to live normal lives. Previous studies similarly found this tendency to discuss 'a normal life' in relation to SCD cures among SCD patients and caregivers[29]. Self-perception of SCD burden appeared to be associated with attitudes towards genetics research and SCD cures. Previous studies found that self-perception of SCD burden played a role in attitudes towards BMT among patients and caregivers in the US. Those who saw their cases as less severe or felt less limited by the disease were less likely to want the procedure[29,30]. As previously described, 4 patients in this study indicated that they had no interest in receiving SCD cures. These patients had more positive self-perception and maintained that they neither felt they were treated differently nor felt limited by the disease.

Many respondents also recognized that SCD cures had some limitations. For example, they considered that older patients or those with progressive damage were less likely to benefit in this way. Those with more severe symptoms were thought more likely to benefit than those with milder cases. Finally, patients, parents and providers wondered how lifestyle or biological factors would influence the cures' effectiveness. These considerations pointed to respondents' attitudes towards health as they considered what factors, such as lifestyle, mindset, biology and one's social or physical environment were most instrumental in determining one's overall health.

Respondents also considered the cures' potential risks. Of the risks, patients, parents and providers described concerns about the chemotherapy involved in BMT and the associated risks of hair loss, infertility and death. Similarly, they worried about the potential side effects of gene-based cures and the use of an HIV derived viral vector in gene therapy. Patients and parents were adamant about not exchanging one disease or illness for another.

Participant attitudes towards genetics research highlighted key ethical issues. The proposed framework highlights these connections **(Figure 4).** Parents, providers and patients all discussed the importance of benefit-sharing in genetics research. Researcher obligation to participants and host communities in LMICs is a widely recognized and discussed ethical standard. The H3Africa benefit sharing model, for example, calls for research groups to ensure equitable benefit sharing with host communities[31]. In this study, participants' views on benefit sharing pointed to a model that prioritized participant benefit, though community benefit was only slightly less favored. Participants in this study described both return of results and concrete, research derived products as the benefits of research participation. Providers believed that researchers were to ensure that participants and their wider communities understood advances made in SCD knowledge through research, and the role they played in the process. All participant groups also maintained that study participants, and to a lesser extent, host communities, were to benefit from the concrete products of research, such as novel SCD treatments. They

believed that this was particularly necessary, given the financial and infrastructural constrains that limited SCD treatment accessibility for most Jamaican patients. As a result, researchers had an obligation to ensure that participants and the wider community benefitted from products derived from research they contributed to.

Participants' attitudes towards autonomy reflected their views on trust and trustworthiness in research and medicine. Providers who more frequently recognized the potential for misuse and misconduct in research tended to favor tighter control through specific or dynamic consent, particularly in terms of biospecimen use and storage. Most providers believed that researchers needed to contact and reconsent participants for the use of their previously donated samples, or explicitly outline study details at the time of sample donation to best respect participant autonomy. Providers were generally cognizant of the harm researchers could cause to participants whether intentionally or unintentionally. Therefore, they saw some restriction as necessary.

Patients on the other hand saw no point to such tight control and favored broad consent in sample use and storage. Some felt researchers could use previously donated samples with some limitation, such as only for SCD specific research. Parents were evenly split between loose and tight control. The H3Africa ethical framework prioritized broad consent in biospecimen use and storage. It emphasized the need for future and secondary sample use to support the goals of the initiative and argued that broad consent would facilitate these goals [32,33]. Other studies found that several LMIC communities found broad

consent to be generally acceptable and more practical than dynamic consent[24,34]. Finally, an H3Africa Ethics consultation found that broad consent was gradually becoming standard throughout many African countries [33]. Caulfield and Murdoch [35] however warned that while broad had grown in popularity, its ethical and legal appropriateness remained contested.

Finally, participants' concerns about invasiveness and loss of privacy reflected the importance they attached to confidentiality in research. Providers, parents and patients emphasized the need to ensure that participant information was sufficiently deidentified and that DNA samples or resulting information could not be linked back to the participant. Buseh, et al. [22], for example, found that among Black African immigrant community leaders, confidentiality was fundamental in whether their people would participate in genomics research and DNA biobanking. Parents and patients in this study similarly maintained that confidentially greatly impacted on their attitudes towards biospecimen collection, storage and use, and data sharing.

Social and cultural issues also explored in this study provide context for participants' attitudes towards genetics research and SCD cures. The final sections of the proposed framework explore the connection between these issues and participants attitudes (**Figure 4**). Socioeconomic challenges emerged as an important factor in patients' and parents' SCD experiences and management. SCD complications such as chronic pain, and leg ulceration resulted in limited education, underemployment and unemployment

among patients. The more severe these complications, the greater the likelihood of facing these limitations[36,37]. As a result, many patients faced financial insecurity and difficulty accessing both conventional and curative SCD treatments which negatively impacted their quality of life. Local health system limitations often exacerbated these issues as patients were generally unable to benefit from free or discounted medications offered through the public health system. Moreover, treatments such as bone marrow transplantation (BMT) were not offered on the island, which required patients to travel abroad to access them. Though some financial assistance was available, it was often limited in the kind and amount of support it could provide. Many participants saw genetics research, and research participation in general as a way to access healthcare. In fact, some participants did describe instances of SCD patients receiving care through affiliation with a study at the SCU. These factors likely helped fuel the diagnostic and therapeutic misconceptions described previously.

Many patients also experienced enacted stigma in many facets of their lives. This included in interpersonal relationships and in the workplace. Providers also described stigma against SCD patients in the healthcare system where patients were often labelled as drug seeking or addicts, and as a result were given lesser quality care. Previous studies demonstrated that Jamaican adults living with SCD faced a moderate amount of stigma[10,36]. Notably, Blake, et al. [10] found that poor socioeconomic status worsened

patients' experiences with stigma. Moreover, those patients with more severe symptoms or outward signs of the disease experienced more extreme health related stigma.

Patients also experienced gender specific stigma. Male patients' masculinity and strength were called into question or dismissed. Female patients experienced stigma related to infertility. Some studies reported lower fecundity among women with the SS phenotype when compared with their AA counterparts[38-40]. They also described high rates of spontaneous abortion, and lower birth weight and gestational age at delivery among those able to conceive[38,39]. Other studies, however, concluded that there was no consensus on relative infertility in women with SCD[41]. Nonetheless, providers and patients in the study reported that many female patients struggled with infertility. Women with SCD also faced an increased risk of maternal mortality[38]. Providers in this study described patients who were highly driven to have children, sometimes at the expense of their health. Many of these women experienced stigma in their romantic relationships and in the wider community.

Notably, however, providers, parents and patients all placed emphasis on decreasing the incidence of children born with SCD. Women in the study therefore also experienced conflict over their desire to have children and the risk of giving birth to a child with sickle cell disease. These women, and other patients, parents and providers, tended to also describe the importance of ensuring that those with SCD chose partners

that did not have the sickle cell trait. Among some women, therefore, the desire to have a child greatly influenced attitudes towards SCD cures and genetics research.

SCD patients experienced significant emotional and mental challenges. Social economic issues and stigmatizing experiences have been found to cause significant psychological challenges in patients[10,11,36]. In this study, much of these effects manifested as internalized or anticipated stigma, and self-limiting behaviors. Most patients seemed to believe consciously or subconsciously that they did not live normal lives due to SCD. These patients described feeling as if they were not normal or described others as normal in contrast to themselves. Some delayed disclosing their SCD disease status for fear of social repercussions, sometimes to the detriment of their wellbeing, and displayed self-limiting behaviors, such as refusing to socialize with peers or pursue romantic relationships.

Providers, patients and parents therefore saw SCD cures as keys to achieving good health and overcoming the socioeconomic and psychosocial challenges patients experienced due to SCD. They believed that in so doing, SCD cures would allow patients to live 'a normal life'. **Figure 4** shows this connection between these challenges and respondents' belief that SCD cures would lead to a normal life for patients.

Faith and spirituality also played a role in coping with the challenges caused by the disease for both patients and parents. These patients and parents relied on their faith for physical, mental and emotional wellbeing. Faith and spirituality also related to

participants' attitudes towards genetics research and SCD cures. A few patients worried about the ramifications of altering genes that they believed God made unique for each person. Faith and spirituality also influenced some patients' and parents' mistrust of researchers and genetics research. However, opposite to that reported by Dennis-Antwi, et al. [20] of religion among some Ghanaian SCD stakeholders, in general, faith and spirituality did not appear to unduly influence or disrupt patients' and parents' health seeking behavior.

Finally, though patients and parents looked to a number of sources such as religion, family and friends, other SCD patients and parents, and the internet for SCD information and influence in SCD related decision making, they most frequently relied on the SCU. Patients and parents generally displayed trust in both SCU researchers and healthcare providers. Providers and researchers at the SCU developed close relationships with parents and patients and acted as major sources of social, financial and emotional support. They were therefore also instrumental in patient and parent coping. Through this relationship, they earned their patients' trust and contributed to their openness to medical research in general.

This trust sometimes manifested as a sense of deference to research and researchers. As previously described, SCD patients and parents tended to place significant trust in researchers and assumed that they worked to their benefit. They often did not consider the ways in which researchers could cause them harm through sample misuse

or research misconduct. As a result, they were willing to leave matters of sample use and storage to researcher discretion. Moreover, some individuals saw themselves as powerless. They believed they could have no control over their samples and appeared to be largely accepting of this fact.

4.1 Implications for policy and practice

The proposed framework described above points to a number of social, ethical and cultural issues researchers in genetics and clinicians must consider in conducting genetics research and introducing gene-based cures to the Jamaican SCD population. Genetics research in low resource settings carries unique ethical implications. Power imbalances are at the basis of many of these implications, particularly those that exist between foreign researchers in genetics and host communities. Factors such as limited education, poor socioeconomic backgrounds and disability often exacerbate these imbalances that can have severe impact on participant autonomy[42]. Participants' misconceptions about the nature and purpose of genetics research and their rights as research participants, as well as undue inducement can perpetuate feelings of powerless in research processes, facilitate participant exploitation and diminish participant autonomy through impaired informed decision making. Close collaboration with local partners such as the SCU will be instrumental in both garnering participant trust and ensuring that their voices are heard, and needs accounted for.

While the SCU will likely play an important role in ensuring patients' autonomy is preserved, greater communication and understanding of research goals and the implications of research participation is also an essential part of this process. Misconceptions about research processes, tendencies to overlook the potential for misuse or misconduct, and reported patient and parent conflation of SCD research and treatment could negatively impact decision making. Moreover, they can create unrealistic expectations among participants and place them at greater risk for harm and exploitation. Improved education and programming focused on genetics research will greatly minimize these risks and further facilitate informed decision making among SCD participants.

Finally, to minimize potential harm, maximize potential benefits and support participant power and autonomy in research, researchers in genetics must work with the local community to develop an explicit and comprehensible benefit sharing model. Researchers in genetics must provide fair benefits to participants and host communities for genetics research to which they contribute either through biospecimen donation or otherwise. Ethics discourse introduces two benefit sharing models for genetics and genomics research, the reasonable availability and fair benefits model[31]. The first mandates the at least partial flow of research derived benefits to the host community while the second allows that the host community negotiate for benefits beyond those derived from research in order to best respect its autonomy[31]. H3Africa argues on behalf

of a third model. It maintains that genetics and genomics research should result in capacity building within host communities[31]. While these study respondents' perspectives point to the first model, researchers must work closely with host communities to ensure that the merits and shortcomings of each model are understood clearly and completely, and if necessary, develop a context specific variant. A model that integrates the reasonable availability model, and the model proposed by H3Africa, for example, may be optimal in the Jamaican setting. This is because treatment accessibility for Jamaican patients is challenged by both financial and infrastructural constrains. Prioritizing research derived benefits and capacity building therefore increases the likelihood that the Jamaican community will achieve the most benefit from research in which they participate, and furthers the overall goal of integrating curative technologies like gene editing and gene therapy into patient SCD care.

Misconceptions and lack of knowledge about SCD cures may point to the need for improved professional and public education and programming, particularly if these options become more readily available. Greater understanding of the risks and benefits associated with these technologies, as well as their limitations will allow for greater health autonomy. Risks such as the chances of transplant failure, death or infertility associated with BMT need to be explicitly detailed. This last is particularly essential given the social consequences of infertility for women with SCD in Jamaica, the cultural or personal expectation of pregnancy, and patients' expressed desires to receive SCD cures in order

to have a child. These factors must also inform genetic counselling, family planning, and education and programming regarding partner selection and screening.

Finally, given the socioeconomic and psychosocial impact of SCD on patients and their families, health systems and governments must develop social and counselling services that assist patients' adjustment as these cures become more readily available in the long term. Particularly given the reported expectation of SCD cures to give patients 'a normal life', successful transition will heavily depend on this social and emotional support.

4.2 Implications for further research

No studies have engaged Jamaicans on issues related to genetics research and cures for SCD. As SCD treatment shifts to focus on more targeted, curative interventions, the need to consider the perspectives of those from communities of high SCD prevalence grows. Further research is urgently needed to build on the perspectives described here. This study engaged adult patients, parents and SCD providers. However, future studies must consider adolescent patients' perspectives. This age group likely faces unique challenges and complications that were overlooked here. This study also identified concerns about invasiveness and exposure through research and how they related to the ethical principle of confidentiality. Studies can explore more deeply the value of privacy in health and family matters in Jamaican culture and its association with genetics research attitudes and participation.

This study also explored patients' willingness to receive SCD cures and identified common characteristics among those who reported no desire to receive them. Further studies are needed to more definitively determine and characterize factors that are associated with SCD cure willingness and hesitancy.

Finally, few studies have looked at SCD patients' experiences with BMT and its aftermath in the US[29,30] Fewer still have looked at SCD patients' experiences with gene-based treatments and tend to focus on clinical outcomes[14]. No studies, however, have looked at the social and psychological effects of receiving SCD cures in Jamaica or other low resource settings. While this is likely due to limited accessibility and therefore the small number of patients that have received these treatments, as health systems and researchers work to make them more readily available, they must examine their potential and actual social and psychological impact on patients and their families.

4.3 Study strengths and limitations

This study used a qualitative approach to examine SCD stakeholder perspectives on genetics research and cures for SCD in Jamaica. It is the first study to engage the Jamaican community on issues relating to genetics research such as biospecimen use and storage. Moreover, it is among the first to explore these issues alongside those related to both approved and developing SCD cures globally. This study also considered a wide range of perspectives, including those from individuals living with SCD, parents of individuals living with SCD, and SCD healthcare providers. The study achieved thematic

saturation. This speaks to the strength of the interview process. Further, more than half of the island's 14 parishes were represented in this study. However, a relatively small sample size suggests that this sample may not have representative of the general population.

Providers were recruited through convenience sampling. Patients and parents on the other hand were selected randomly from patient files at the SCU. Purposive sampling was employed to ensure that male patients and parents were also recruited into the study. Convenience sampling potentially introduced bias into the study, limiting the range of perspectives to those provided by providers who were most available or open to participate.

Due to the constrains of the COVID-19 pandemic, this study faced unique limitations. This study was conducted remotely over the Zoom videoconferencing platform. This particularly affected patient and parents as only those able to access the required hardware and software and a stable internet connection were able to participate in the study. This created another source of bias, potentially systematically excluding the unique perspectives of patients and parents with certain sociodemographic characteristics.

Remote interviews also led to increased distraction as participants conducted interviews from home. Noisy environments and poor internet connections led to problems with audio and distractions that likely negatively affected the quality of the

interviews. Key perspectives and insights therefore may have been lost due to these distractions.

Finally, discussing technical concepts regarding genetics research and gene-based SCD cures created certain limitations. Some of the perspectives provided here may have largely been influenced by misunderstanding or confusion about these topics. This could limit the validity and applicability of some of the study's findings. However, this also creates a basis for future, more robust qualitative and quantitative studies.

4.4 Researcher Positionality

The researcher is a Jamaican female student engaging Jamaican stakeholders. In some respects, this affords the researcher the position of an insider. However, key differences in other sociodemographic factors such as education and gender, as well as potential power imbalances between researcher and participant moderate this position. Differences in knowledge about SCD research and cures, for example, may have precipitated an imbalance between the researcher and parents and patients, that may have negatively impacted the quality of the interview.

An insider positionality likely created unique strengths and limitations. The researcher may have been better able to build rapport and allow participants to feel more at ease in the research process, and free to express themselves more naturally. For many, Jamaican creole or patois is the dialect of choice. An insider positionality, therefore, allowed participants to speak in a way that was more comfortable for them. Furthermore,

this positionality enabled the researcher to identify important linguistic details and identify key context clues. However, the insider positionality potentially created bias through the interview and analysis processes, particularly due to the nature of qualitative research in which the researcher also acts as an instrument. Viewing and interpreting participant responses through the lens of an insider may have impacted the validity of the conclusions drawn.

5. Conclusion

Despite some skepticism and reluctance, patients, parents and providers were optimistic about genetics research. They hoped it would lead to improved SCD knowledge and treatment options that could minimize or entirely eliminate challenges caused by SCD. Participant attitudes towards genetics research pointed towards important ethical guidelines researchers in genetics must consider. These guidelines include benefit-sharing models that prioritize the supply of research derived products to research participants and host communities, as well as those that govern autonomy and confidentiality. Patients, parents and providers also viewed SCD cures favorably, though there was concern about the risks and questions about whether and which patients would benefit from these cures. Respondents also described challenges including frequent pain, leg ulceration, and infertility and the resulting socioeconomic limitations, stigma, and negative emotional and mental health effects. Respondents generally believed these

challenges limited patients' ability to live a 'normal life' and therefore saw SCD cures as the key to a 'normal life'

Appendix A

Provider Consent Form to Participate in Research

Introduction

This study is being conducted by Krystin Jones of the Duke Global Health institute at Duke University and is in partial fulfillment of her Masters in Global Health degree. Her supervisor in Jamaica is Dr. Monika Asnani from the Sickle Cell Unit at the UWI.

Why is this study being done?

Sickle cell disease is a genetic disease. There have been great advances in genetics research and gene-based cures for sickle cell disease. Globally, there has been much research examining the genetic aspects of origin and course of the disease, but less research on the views and experiences of the research participants, other people living with sickle cell disease, and other stakeholders such as healthcare providers managing sickle cell disease concerning these advances. Moreover, while bone marrow transplantation remains the only curative technique currently in clinical practice, other gene-based techniques such as gene transfer and gene editing are currently undergoing clinical trials for use in sickle cell disease and show great promise for application in African and Caribbean communities. It is essential that the implementation of genetics research and emerging technologies take into account the needs and the values of these communities.

The overall purpose of this study is to the explore perspectives on genetics research and cures for sickle cell disease with the aim of obtaining an understanding of the ethical, social and cultural factors that relate to these perspectives.

What will I be asked to do?

You are being asked to participate in this study because you are a health care provider who treats individuals living with sickle cell disease. If you choose to participate in this study, you will be asked to sign and date this consent form, and you will receive a copy of the signed form. You will then be asked to take part in an in-depth interview that will be conducted using secure remote conferencing tool such as Zoom or WebEx. We will ask about your professional experiences with sickle cell disease and your opinions on genetics research, treatments and cures for sickle cell disease. With your permission, the interview will be digitally recorded and captured through hand-written notes. All recordings will be destroyed after they have been transcribed.

How long will I be in the study?

We expect your participation in the study to last between 1 and 1.5 hours.

What are the risks and inconveniences and benefits of this study?

No physical harm to you is anticipated by your participation in the study. If you experience any emotional discomfort, we will refer you to speak with our social worker at the Sickle Cell Unit. You may refuse to answer any question at any time, to take breaks from the interview as needed, or to withdraw from the study all together.

There are no immediate benefits to you for participating but hopefully, this study will provide further insights into genetics and genomics research and help inform a framework with which responsible and ethical genetic research may be conducted and with which new technologies may be implemented in these communities in relation to sickle cell disease.

Confidentiality:

We will keep this signed consent form in a secure location separate from your data. A unique code number will be assigned to all data we collect from you. All data will be stored in Duke University's secure cloud-platform, Duke Box. While the data we collect from this study may be presented at scientific meetings or published in a scientific journal, your identity will not be revealed.

Collected data may be made public or used for future research purposes, your identity will always remain confidential. The study results and documentation will be retained for at least six years after the study is completed.

Voluntary nature of participation:

Participation in this study is completely voluntary. Once the study has begun, you are free to withdraw at any time. Refusal to participate or withdraw from the study will in no way affect your current working relationship at the Sickle Cell Unit. Upon withdrawal, no new information will be collected, with the exception of that which is needed to keep track of your withdrawal.

Whom do I call if I have questions or problems?

If you have any questions about the study, you may contact Krystin Jones by telephone at (312)-292-0273, or by email at krystin.d.jones@duke.edu. You may also contact Dr. Monika Asnani, Sickle Cell Unit, CAIHR, UWI by telephone at (876)-927-2471 or email at monika.parshadasnani@uwimona.edu.jm. For questions about your rights as a participant in this research study, please contact Dr. Gillian Wharfe, Chair, Faculty of Medical Sciences, University of the West Indies, Mona, Kgn 7 by telephone at (876) 970-4892 or by e-mail at mcrec@uwimona.edu.jm.

Statement of Consent

Do you have any questions about the study? [ ] Yes [ ] No

Would you like us to email or mail you a copy of this consent form? [ ] Yes [ ] No

Will you participate in this research? [ ] Yes [ ] No

I prefer that the interview not be recorded [ ] Yes [ ] No

I am willing to participate in video call [ ] Yes [ ] No

[If participant responds yes, then complete signatures below; otherwise respond "thank you for your time" and end telephone conversation.]

Name of Respondent:_____

Signature of Respondent: _____ DATE:_____

Name of Researcher:_____

Signature of Respondent:_____ Date:_____

Signature of Independent Witness: _____

Patient Consent Form to Participate in Research

Introduction

This study is being conducted by Krystin Jones of the Global Health Institute Duke University in partial fulfilment of the Master of Science in Global Health degree under the supervision of Dr. Monika Asnani from the Sickle Cell Unit at the UWI.

Why is this study being done?

Sickle cell disease is a genetic disease which results from being born with two genes for sickle hemoglobin. Genes are what we inherit from both our parents and that give us many of our characteristics, for example the colour of our eyes or our height. All through the world there is much research to understand genes and how they interact with our environment, such as the climate of the place where we live or our social circumstances such as educational level, to affect how healthy we are or how illness behaves in our bodies.

We know bone marrow transplant is a cure for sickle cell disease, but we also know that there is now research to study if we can alter our genes in any way to cure us of sickle cell disease. However, there is not much research on the views and experiences of people living with sickle cell disease, their family members or even among their healthcare providers about these efforts to cure sickle cell disease. It is important to understand this so researchers can take into account the needs and the values of these communities.

The overall purpose of this study is to the explore perspectives on genetics research and cures for sickle cell disease with the aim of obtaining an understanding of the ethical, social and cultural factors that relate to these perspectives.

What will I be asked to do?

If you choose to participate in this study, you will be asked to read, sign and date this consent form, and you will receive a copy of the signed form. You will then be asked to take part in an in-depth interview which will last between 1 and 1.5 hours. You will be asked questions about your own experience with sickle cell disease, your opinion on participating in genetics research, and how your information should be used. Other questions will look at your understanding of cures that are available and your views about those cures.

This interview will be done using a video call. With your permission, the interview will be digitally recorded and captured through hand-written notes, so that we can make accurate transcripts. All recordings will be destroyed after they have been transcribed.

How long will I be in the study?

We expect your participation in the study to last between 1 and 1.5 hours.

What are the risks and inconveniences of this study?

There are no expected risks to you from participating in this research study. However, topics discussed during the interview may cause some emotional discomfort. You are able to refuse to answer any question at any time, to take breaks from the interview as needed, or to withdraw from the study all together. At the end of the interview, you may choose to receive counselling from the social worker attached to the Sickle Cell Unit.

Benefits and Compensation:

Should you choose to participate in this study, you will receive a compensation of $1000 JMD for your time. If you have to buy internet service for the duration of the interview, we will also compensate you for that cost.

There are no immediate health benefits to you by participating in this study but hopefully we will get a deeper understanding of your needs and views regarding genetic research and cures for sickle cell disease. This will help us and other researchers to design better studies that meet your needs.

Confidentiality:

We will keep this signed consent form in a secure location separate from your data. A unique code number will be assigned to all data we collect from you. All data will be stored in Duke University's secure cloud-platform, Duke Box. While the data we collect from this study may be presented at scientific meetings or published in a scientific journal, your identity will not be revealed.

Collected data may be made public or used for future research purposes, your identity will always remain confidential. The study results and documentation will be retained for at least six years after the study is completed.

Voluntary nature of participation:

Participation in this study is completely voluntary. Once the study has begun, you are free to withdraw at any time. Refusal to participate or withdraw from the study will in no way affect your relationship with clinic staff and health care providers, or your access to the care to which you are entitled at the Sickle Cell Clinic. Upon withdrawal, no new information will be collected, with the exception of that which is needed to keep track of your withdrawal.

Whom do I call if I have questions or problems?

If you have any questions about the study, you may contact Krystin Jones by telephone at (312)-292-0273, or by email at krystin.d.jones@duke.edu. You may also contact Dr. Monika

Asnani, Sickle Cell Unit, CAIHR, UWI by telephone at (876)-927-2471 or email at monika.parshadasnani@uwimona.edu.jm. For questions about your rights as a participant in this research study, please contact Dr. Gillian Wharfe, Chair, Faculty of Medical Sciences, University of the West Indies, Mona, Kgn 7 by telephone at (876) 970-4892 or by e-mail at mcrec@uwimona.edu.jm.

Statement of Consent

Do you have any questions about the study? [ ] Yes [ ] No

Would you like us to email or mail you a copy of this consent form? [ ] Yes [ ] No

Will you participate in this research? [ ] Yes [ ] No

I prefer that the interview not be recorded [ ] Yes [ ] No

I am willing to participate in video call [ ] Yes [ ] No

[If participant responds yes to participating, then complete signatures below; otherwise respond "thank you for your time" and end telephone conversation.]

Name of Respondent:_____

Signature of Respondent: _____ DATE:_____

Name of Researcher:_____

Signature of Respondent:_____ Date:_____

Signature of Independent Witness :_____

Parent Consent Form to Participate in Research

Introduction

This study is being conducted by Krystin Jones of the Global Health Institute Duke University in partial fulfilment of the Master of Science in Global Health degree under the supervision of Dr. Monika Asnani of the Sickle Cell Unit of the UWI.

Why is this study being done?

Sickle cell disease is a genetic disease which means it is a result of a being born with two genes for sickle cell disease. Genes are what we inherit from both our parents and that give us all our attributes, for example the colour of our eyes or our height. All through the world there is much research to understand genes and how our environment, such as the climate of the place where we live or our social circumstances such as educational level, might affect how healthy we are or how our illness behaves in our body.

We know bone marrow transplant is a cure for sickle cell disease, but we also know that there is now research to study if we can alter our genes in any way to cure us of sickle cell disease. There is however not much research on the views and experiences of people living with sickle cell disease, their family members or even among their healthcare providers. It is important to understand this so researchers can take into account the needs and the values of these communities.

The overall purpose of this study is to the explore perspectives on genetics research and cures for sickle cell disease with the aim of obtaining an understanding of the ethical, social and cultural factors that relate to these perspectives.

What will I be asked to do?

If you choose to participate in this study, you will be asked to sign and date this consent form, and you will receive a copy of the signed form. You will then be asked to take part

in an in-depth interview which will last between 1 and 1.5 hours. You will be asked questions about your and your child's experiences with sickle cell disease, your opinion on participating in genetics research, and how your information should be used. Other questions will look at your understanding of cures that are available and your views about those cures.

This interview will be done using a video call. With your permission, the interview will be digitally recorded and captured through hand-written notes, so that we can make accurate transcripts. All recordings will be destroyed after they have been transcribed.

How long will I be in the study?

We expect your participation in the study to last between 1 and 1.5 hours.

What are the risks and inconveniences and benefits of this study?

There are no expected risks to you from participating in this research study. However, topics discussed during the interview may cause some emotional discomfort. You are able to refuse to answer any question at any time, to take breaks from the interview as needed, or to withdraw from the study all together. At the end of the interview, you may choose to receive counselling from the social worker attached to the Sickle Cell Unit

Compensation:

Should you choose to participate in this study, you will receive a compensation of $1000 JMD for your time. If you have to buy internet service for the duration of the interview, we will also compensate you for that cost.

Confidentiality:

We will keep this signed consent form in a secure location separate from your data. A unique code number will be assigned to all data we collect from you. All data will be stored in Duke University's secure cloud-platform, Duke Box. While the data we collect from this study may be presented at scientific meetings or published in a scientific journal, your identity will not be revealed.

Collected data may be made public or used for future research purposes, your identity will always remain confidential. The study results and documentation will be retained for at least six years after the study is completed.

Voluntary nature of participation:

Participation in this study is completely voluntary. Once the study has begun, you are free to withdraw at any time. Refusal to participate or withdraw from the study will in no way affect your or your child's relationship with clinic staff and health care providers, or your child's access to the care to which he or she is entitled at the Sickle Cell Clinic. Upon withdrawal, no new information will be collected, with the exception of that which is needed to keep track of your withdrawal.

Whom do I call if I have questions or problems?

If you have any questions about the study, you may contact Krystin Jones by telephone at (312)-292-0273, or by email at krystin.d.jones@duke.edu. You may also contact Dr. Monika Asnani, Sickle Cell Unit, CAIHR, UWI by telephone at (876)-927-2471 or email at monika.parshadasnani@uwimona.edu.jm. For questions about your rights as a participant in this research study, please contact Dr. Gillian Wharfe, Chair, Faculty of Medical Sciences, University of the West Indies, Mona, Kgn 7 by telephone at (876) 970-4892 or by e-mail at mcrec@uwimona.edu.jm.

Statement of Consent

Do you have any questions about the study? [ ] Yes [ ] No

Would you like us to email or mail you a copy of this consent form? [ ] Yes [ ] No

Will you participate in this research? [ ] Yes [ ] No

I prefer that the interview not be recorded [ ] Yes [ ] No

I am willing to participate in video call [ ] Yes [ ] No

[If participant responds yes, then complete signatures below; otherwise respond "thank you for your time" and end telephone conversation.]

Name of Respondent:_____

Signature of Respondent: _____ DATE:_____

Name of Researcher:_____

Signature of Respondent:_____

Date:_____

Signature of Independent Witness :_____

Healthcare Providers Demographics Survey

To be completed by interviewer

Sex

M/F

Introduction

(Start recording) Hello, my name is Krystin Jones and I am studying for a Master's Degree at Duke University in the USA. This research study is in partial completion of this degree. Today, I want to talk to you about your perspectives on genetics research and cures for sickle cell disease. I will be asking questions about your personal and professional experiences with the disease, your thoughts on current treatments for the disease, your opinions on genetics research, and what you think about cures for sickle cell disease. All of your responses are valuable, so please feel free to share whatever you think is important.

Do you have any questions before we begin?

Demographic Information

I would like to start by gathering some demographic information about you

How old are you?

Where were you born?

With what racial or ethnic group do you identify?

What is your highest Education Level?

Primary (Grades 1-6)
Secondary (7-11)
Post-Secondary
College or University
Post University or Professional Degree

Can you confirm that your official job title is _____?

What is your religion?

Does anyone in your biological family have sickle cell disease?

Biological Mother
Biological Father
Brother
Sister

Healthcare Provider In-depth Interview Guide		
Question	Focus of Question Related to Methods or study Aims	Probes
Part 1- For this first set of questions, I want to get a general sense of your work with sickle cell disease		
When did you start working with patients with SCD?	To build rapport	What does a normal day working with SCD patients look like for you?
Where did your interest in working with patients with sickle cell disease come from?	To build rapport; to gain a sense of participant's interests and expertise	Can you describe your academic and professional journey that led you here? Can you tell me about any personal experiences you have had with sickle cell disease?
Tell me about some of the patients you see most frequently	To build rapport; to ease the participant into talking about sickle cell disease experiences	Can you tell me about any cases that have stood out to you? What are some of the main challenges your patients face? (Physically and emotionally)
What kind of self-management techniques are you familiar with among patients?	To get a better understanding of participant's professional experiences with the disease	What are some techniques that you have recommended to your patients?
What are some of ways that your patients cope emotionally with having the disease?	To get a better understanding of participant's professional experiences with the disease	Are there any that you find concerning? What do you tend to recommend?

Part 2- Genetics and genomics research have attracted a lot of attention in recent years. For this next set of questions, I want to talk with you about your understanding of genetics research and some of the issues that might be associated with it. These include sample collection, storage and sharing. I also would like to talk about why you think some people might take part in these kinds of research. This is all still quite new, and most people don't know much about it. So again, it's okay if you don't know the answers, and if something isn't clear, feel free to let me know and ask any questions you need to. *-These questions are adapted from the Sickle Cell Disease Genomics Network in Africa's (SickleGenAfrica) community engagement format and survey*		
To start, what do you think of when you hear the term genetics research?	To get an understanding of how participant's conceptualize genetics research	Are there any personal stories that come to mind? Are there any professional ones? What is the purpose of genetics research?
If unfamiliar: Genetics research studies the roles your genes play in certain traits, like the color of your eyes, the color of your hair development of certain diseases.		
How do feel about researchers having access to all the information stored in a participant's genes?	To get an understanding of participant's perspectives on genetics research	Do you have any concerns about this?
Why do you think a person (with sickle cell disease) might participate in this kind of research?		What are some reasons people would not want to participate?
Do you think that it's important for Jamaicans to participate in research?		Who do you think should benefit first from results that come out of genetics research?
How would you decide about whether you would recommend for a patient to participate in genetics research?	To get an understanding of participant's perspectives on genetics research	Are there cases where you would absolutely recommend against it?

In your experience, how do individuals living with sickle cell disease make decisions regarding the management or treatment of their illness?	To investigate ideas of autonomy, informed consent and shared decision making	Who do you find has the most influence in this respect?
Hospitals and Researchers collect samples like blood, hair, saliva, urine for genetics research.		
What kind of samples do you think a person with sickle cell disease would feel most comfortable providing?	To investigate perspectives on sample collection, management and storage	Which ones would you think of as off limits? How do you think the kind of sample a study collects influences a person's decision to participate in genetics research?
Researchers may also store samples for many years in biobanks. In the future, different researchers or organizations like pharmaceutical companies, insurance companies or even governments, may be able to go to these biobanks and take out these samples and use them in their own research or in other ways		
What do you think about storing samples in biobanks?		Do you have any concerns about this? What rights should participants have over these samples? Control, benefits?
What do you think about storing samples from Jamaican participants in biobanks located overseas?		How do you think this could impact the Jamaican participant's rights?
Should there be a limit for how long samples can be stored?	To investigate perspectives on sample collection, manage and storage	What do you think about samples being stored indefinitely?

How do you feel about researchers or other organizations in Jamaica or other countries being able to get and use samples stored in a biobank?	To investigate perspectives on sample collection, manage and storage	Is there any group or organization that you feel should not be able to access participant's sample?
How do you feel about researchers using these samples in many studies or in many different ways?	To investigate perspectives on sample collection, manage and storage	Are there things you wouldn't want these samples to be used for?
Part 3- For this third set of questions, I would like us to discuss how you understand and what you think about existing treatments and the potential cures for sickle cell disease.		
What does good health mean to you?	To investigate the ways in which participants think about health	Do you think anyone can achieve good health? What are some of the ways you recommend that someone with SCD try to achieve good health?
There is a great deal of variation in symptoms and illness severity in people with sickle cell disease. What do you think accounts for this?	To investigate the ways in which participants think about health	Do you think that social or interpersonal factors can play a role in this?
Can you tell me about the current treatment options available for sickle cell disease?	To assess participants understanding of and perspectives on treatments for sickle cell disease	Can you tell me about the main side effects of these treatments? What have your patients told you about their ability to keep up with these treatments?

How would you define a cure for sickle cell disease?	To investigate participants' understanding of and interest in a cure for sickle cell disease	What impact do you think it could have on a patient's life? What challenges do you think would remain? Do you think there would be any new challenges that could arise?
Currently, bone marrow transplantation is the only approved cure for sickle cell disease.		
Do you know of any patients that have undergone bone marrow transplantation?	To investigate participant's experiences with sickle cell disease	If yes, what was the patient's experience like? If no, have you heard anything about it or come across it in any other way?
There are 2 cures for SCD that are currently being developed called gene editing and gene transfer therapy that change your genes or how they are expressed.		
What do you think about treatments that make changes to a person's genes?		What are some concerns you may have?
Would you recommend these treatments to your patients while they are still in clinical trials?	To investigate participants understanding of and interest in a cure for sickle cell disease	What are some things you think a patient would need to know about these kinds of treatments?
To your knowledge, is there any infrastructure in place that would allow Jamaican patients to benefit from these treatments?	To investigate participants understanding of and interest in a cure for sickle cell disease	Do you think Jamaican patients are likely to benefit from these cures?

		In your opinion, what is the main factor that would determine whether Jamaican patients would benefit from these cures? Do you think these cures would make much difference in the Jamaican setting? Why or Why not?
Part 4- This final set of questions go into how people with sickle cell disease are treated in your community.		
In your experience, are people with sickle cell disease treated differently from other people in Jamaica	To explore the treatment of individuals with sickle cell disease	Can you tell me of any instances in which you witnessed a person with sickle cell disease being treated differently or unfairly because of the disease? Can you tell me of any instances in which you witnessed a parent being treated differently? What are some of the things that people say about sickle cell disease?
What if any systems are put in place to protect people living with sickle cell disease from discrimination or other kinds mistreatment?	To further explore the treatment of individuals with sickle cell disease	Do you think these systems in Jamaica are sufficient?

Would you say that most of your patients with SCD come in with an adequate understanding of the disease?	To explore knowledge of SCD among patients	In your experience, how do people typically learn about sickle cell disease? What misconceptions do you most often encounter?
What, if anything, have you told your patients about disclosure of their sickle cell disease status?	To explore the treatment of individuals with sickle cell disease	What role do you think a support system plays in SCD management? What, in your experience, is the largest source of support for individuals living with SCD?
Finally, do you have any questions or anything else you'd like to add? *Thank you for taking the time to speak with me today*		

Patient Demographics Survey

To be completed by interviewer
Sex
M/F

Introduction
Hello, my name is Krystin Jones and I am studying for a Master's Degree at Duke University in the USA. This research study is important for me to complete this degree. Today, I want to talk to you about your perspectives on genetics research and cures for sickle cell disease. I will be asking questions about your personal experiences with the disease, your thoughts on current treatments for the disease, your opinions on genetics research, and what you think about cures for sickle cell disease. All of your responses are valuable, so please feel free to share whatever you think is important.

Do you have any questions before we begin?

Demographic Information
I would like to start by gathering some demographic information about you

How old are you?

Where were you born?

With what racial or ethnic group do you identify?

What is your highest Education Level
- Primary (Grades 1-6)
- Secondary (7-11)
- Post Secondary
- College or University
- Post University or Professional Degree

What is your occupation?

What is your religion?

Does anyone else in your biological family (apart from you) have sickle cell disease?
- Biological Mother
- Biological Father
- Brother
- Sister

Patient In-Depth Interview Guide		
Question	Focus of Question Related to Methods or study Aims	Probes
Part 1- First, I want to get a general sense of your experiences with sickle cell disease, and here at the sickle cell unit. Just answer these questions as fully as you can and if you're not sure about a question, please don't hesitate to ask for clarification.		
When did you first learn you had sickle cell disease?	To get a better understanding of participant's personal experiences with the disease	How did you learn about it? How was your sickle cell disease explained to you for the first time? How does a person get sickle cell disease? Are there any interesting things you've heard about how a person gets sickle cell disease? Do you remember how you felt in that moment? Did you feel supported?
Please tell me about some of your symptoms	To get a better understanding of participant's personal experiences with the disease	Can you tell me how you've managed these symptoms? How did you come to do these things? What's worked? What hasn't?
Tell me about when you first started attending the sickle cell unit.	To build rapport; to ease the participant into talking about sickle cell disease experiences	How often do you come in to the sickle cell unit? What are some of the services you receive here?

Where do you turn to for information about sickle cell disease?		
Part 2- For this next set of questions, I want to talk with you about your understanding of genetics research and some of the issues that might be associated with it. These include sample collection, storage and sharing. I also would like to talk about why you think some people might take part in these kinds of research. This is all still quite new, and most people don't know much about it. So again, it's okay if you don't know the answers, and if something isn't clear, feel free to let me know and ask any questions you need to. *-These questions are adapted from the Sickle Cell Disease Genomics Network in Africa's (SickleGenAfrica) community engagement format and survey*		
To start, what do you think of when you hear the term genetics research?	To get an understanding of how participant's conceptualize genetics research	Are there any personal stories that come to mind?
If unfamiliar: Genetics research studies the roles your genes play in certain traits, like the color of your eyes, the color of your hair development of certain diseases. Blood relatives often share genes, which explains why parents and their children may look alike or why family members may have certain diseases in common.		
Your genes store information about your body. How do you feel about researchers having access to that information?	To investigate perspectives on sample collection, management and storage	Would you want researchers to contact you to tell you about other things they might find in your genes? Why (not)?
Why do you think a person (with sickle cell disease) might participate in this kind of research?	To get an understanding of participant's perspectives on genetics research	What are some of the things that could keep people from participating in this kind of research?

What are some reasons that you would participate in genetics research?	To investigate ideas of autonomy, informed consent and shared decision making	Is there anyone who you normally turn to for advice about how to manage your illness? Would they influence your decision to participate in genetics research? What are some concerns you would have about participating in genetics research?
Sometimes, the results of genetics research may not be available for a while. Would you want to participate in this research if the results would not be available for many years?	To investigate motivations perspectives on participation in genetics research	Who do you think should benefit from research you participate in?
Hospitals and Researchers collect samples like blood, hair, saliva, urine for genetics research.		
If you were to participate in this kind of research, what kinds of samples would you feel most comfortable providing?	To investigate perspectives on sample collection, management and storage	Why? Which ones would you think of as off limits? Would the kind of sample a study collects influence your decision to participate in genetics research?

Researchers may also store samples for many years in places called biobanks. A biobank is a bank where, instead of money, it stores people's biological samples. In the future, different researchers or organizations like pharmaceutical companies that make medicines, insurance companies or even governments, may be able to go to these biobanks and take out these samples (like taking out money at a bank) and use them in their own research or in other ways		
How do you feel about your samples being stored in a biobank?	To investigate perspectives on sample collection, management and storage	Are there certain rights or powers you would want to have over these samples? Control? Benefits?
Would you want there to be a limit for how long your samples are stored?	To investigate perspectives on sample collection, management and storage	Why did you suggest that amount of time? What should be done with them after this time? Should researchers inform participants when they destroy these samples?
These biobanks may be overseas, like in the US or the UK. How do you feel about your samples being stored in a different country?	To investigate perspectives on sample collection, management and storage	How do you think this could affect you? Do you think it would affect your rights as a donor? Do you think it would affect your ability to get any benefits that may come from the research? Do you think it would affect you being able to have a say in what happened with the samples?

How do you feel about researchers or other organizations in Jamaica or other countries being able to get and use samples stored in a biobank?	To investigate perspectives on sample collection, management and storage	Is there any person or group who you would not want to be able to use your samples?
How do you feel about researchers using these samples in many studies or in many different ways?	To investigate perspectives on sample collection, management and storage	Are there things you wouldn't want these samples to be used for?
Part 3- For this third set of questions, I would like us to discuss how you understand and what you think about existing treatments and the potential cures for sickle cell disease.		
What does good health mean to you?	To investigate the ways in which participants think about health	Do you think anyone can achieve good health? What are some of the ways you try to have good health?
What are some of the treatment options for sickle cell disease that you know of?	To assess participants understanding of treatments for sickle cell disease	How many of these have you tried? Tell me about some of the ones you've preferred Tell me about some of the ones you didn't try or prefer
Can you tell me about some of your experiences with the	To investigate participants experiences with the available treatment options.	How do you keep up with these treatments?

treatments you currently receive?		What are some of the challenges you face?
How would you describe a cure for sickle cell disease?	To investigate participants understanding of and interest in a cure for sickle cell disease	What would it do? What impact do you think it would have on your life? Would you be interested in having access to a cure? Why (Not)?
Currently, bone marrow transplantation is the only approved cure for sickle cell disease. Bone marrow makes blood in the body. A donor gives his or her healthy bone marrow to the patient so that he or she can make healthy blood cells.		
Have you heard about bone marrow transplantation?	To investigate understanding of and perspectives on existing cures for SCD	Is it something that you want to do? Do you think that it's something that you could have access to? Why?
There are 2 cures for SCD that are currently being developed called gene editing and gene transfer therapy. They make changes to your genes that allow your body to make blood cells that don't sickle. They are still being tested in research studies to see how they how they work on people with sickle cell disease.		
What do you think about treatments that could change your genes?	To investigate participants understanding of and interest in a cure for sickle cell disease	What are some concerns you may have? What are some things you would want to know about these kinds of treatments?

Would you want to be a part of the research that tests these cures?	To investigate participants understanding of and interest in a cure for sickle cell disease	Why or why not?
Would you want to have these kinds of treatments?	To investigate participants understanding of and interest in a cure for sickle cell disease	Why or why not? Do you think that there are resources in Jamaica for people to get these cures?
Part 4- This final set of questions go into how people with sickle cell disease are treated in your community.		
Do you feel you are treated differently as someone with sickle cell disease?	To explore the treatment of parents of individuals with sickle cell disease	In what ways? Positive or negative? What are some of the things people say? How do you feel about this? Do you believe any of them are true?
Tell me about how you've decided to tell people about your sickle cell disease status?	To explore the treatment of individuals with sickle cell disease	Are there any times when you've done this that have stood out to you? How did they react? Afterwards, how did you feel about your decision?
How does sickle cell disease affect different aspects of your life?	To investigate the participant's perception of sickle cell disease in relation to social and cultural contexts	Does it limit you in any way? Do you think that there are any opportunities that SCD has caused you to miss out on? How have you tried to overcome these limits?

Do you have a support system (family, friends, religious groups)?		If yes, what role do they play in your coping?
How do you imagine your future?	Broad question to investigate the participant's outlook with sickle cell disease; coping	
Finally, do you have any questions or anything else you'd like to add? *Thank you again for taking the time to talk with me today*		

Parents Demographics Survey

To be completed by interviewer
Sex
M/F

Introduction
Hello, my name is Krystin Jones and I am studying for a Master's Degree at Duke University in the USA. This research study is important for me to complete this degree. Today, I want to talk to you about your perspectives on genetics research and cures for sickle cell disease. I will be asking questions about your personal experiences with the disease, your thoughts on current treatments for the disease, your opinions on genetics research, and what you think about cures for sickle cell disease. All of your responses are valuable, so please feel free to share whatever you think is important.

Do you have any questions before we begin?

Demographic Information
I would like to start by gathering some demographic information about you

How old are you?

Where were you born?

With what racial or ethnic group do you identify?

What is your highest Education Level
Primary (Grades 1-6)
Secondary (7-11)
Post Secondary
College or University
Post University or Professional Degree

What is your occupation?

What is your religion?

Does anyone else in your biological family (apart from your child) have sickle cell disease?
Biological Mother
Biological Father
Brother
Sister

Parent In-depth Interview Guide		
Question	**Focus of Question Related to Methods or study Aims**	**Probes**
Part 1- First, I want to get a general sense of your and your child's experiences with sickle cell disease, and here at the sickle cell unit. Just answer these questions as fully as you can and if you're not sure about a question, please don't hesitate to ask for clarification.		
What year was your child born?	To build rapport; to ease the participant into talking about sickle cell disease experiences	
Tell me about when you first learned your child had sickle cell disease?	To build rapport; to ease the participant into talking about sickle cell disease experiences	How did you feel about how the information was presented to you? Was it clear? How does a person get sickle cell disease? Did you feel supported in that moment?
Have you told your child that he or she has sickle cell disease?	To get a better understanding of participant's personal experiences with the disease	If no, why not? Have you decided when you will tell your child? If yes, can you tell me about how you made the decision. Do you remember how you felt in that moment? How did your child react? Did he/she understand?
Please tell me about some of your child's symptoms	To get a better understanding of participant's personal experiences with the disease	Can you tell me how you've managed these symptoms? How did you come to do these things?

		What's worked? What hasn't?
When did you first begin to take your child to the sickle cell clinic?	To build rapport; to ease the participant into talking about sickle cell disease experiences	How often do you come into to the sickle cell clinic? What are some of the services you receive here?
Part 2- For this next set of questions, I want to talk with you about your understanding of genetics research and some of the issues that might be associated with it. These include sample collection, storage and sharing. I also would like to talk about why you think some people might take part in these kinds of research. This is all still quite new, and most people don't know much about it. So again, it's okay if you don't know the answers, and if something isn't clear, feel free to let me know and ask any questions you need to. ***-These questions are adapted from the Sickle Cell Disease Genomics Network in Africa's (SickleGenAfrica) community engagement format and survey***		
To start, what do you think of when you hear the term genetics research?	To get an understanding of how participant's conceptualize genetics research	Are there any personal stories that come to mind?
If unfamiliar: Genetics research studies the roles your genes play in certain traits, like the color of your eyes, the color of your hair development of certain diseases. Blood relatives often share genes, which explains why parents and their children may look alike or why family members may have certain diseases in common.		
Your genes store information about your body, your relatives or your heritage/ where you come from. How do you feel about researchers having access to this information?		Would you want researchers to contact you to tell you about other things they might find in your child's genes?
Why do you think a person (with sickle cell disease) might participate in this kind of research?	To get an understanding of participant's perspectives on genetics research	What are some of the things that could keep people from participating in this kind of research?

What are some reasons that would cause you to allow your child to participate in genetics research?	To investigate ideas of autonomy, informed consent and shared decision making	Is there anyone that you usually go to for advice on taking care of your child? Would they influence your decision to participate in genetics research? What are some concerns you would have about your child participating in genetics research?
Sometimes, the results of genetics research may not be available for a while. Would you allow your child to participate in this research if the results would not be available for many years?	To investigate motivations perspectives on participation in genetics research	Who do you think should benefit from research your child participates in?
Hospitals and Researchers collect samples like blood, hair, saliva, urine for genetics research.		
If your child were to participate in this kind of research, what kinds of samples would you feel most comfortable with your child providing?	To investigate perspectives on sample collection, management and storage	Why? Which ones would you think of as off limits? Would the kind of sample a study collects influence your decision to allow your child to participate in genetics research?
Researchers may also store samples for many years in places called biobanks. A biobank is a bank where, instead of money, it stores people's biological samples. In the future, different researchers or organizations like pharmaceutical companies that make medicines, insurance companies or even governments, may be able to go to these biobanks and take out these samples (like taking out money at a bank) and use them in their own research or in other ways		

How do you feel about your child's samples being stored in a biobank?	To investigate perspectives on sample collection, management and storage	Are there certain rights or powers you would want to have over these samples? Control? Benefits?
Would you want there to be a time limit for how long your child's sample can stay in the biobank?		What should be done with them after this time?
These biobanks may be overseas, like in the US or the UK. How do you feel about your child's samples being stored in a different country?	To investigate perspectives on sample collection, management and storage	How do you think this could affect you or your child? Do you think it would affect your or child's rights as a donor? Do you think it would affect your ability to get any benefits that may come from the research? Do you think it would affect you being able to have a say in what happened with the samples?
How do you feel about researchers or other organizations in Jamaica or other countries being able to get and use samples stored in a biobank?	To investigate perspectives on sample collection, management and storage	Is there any group that you would not want to be able to get or use your child's sample?

How do you feel about researchers using these samples in many studies or in many different ways?	To investigate perspectives on sample collection, management and storage	Are there things you wouldn't want these samples to be used for?
Part 3- For this third set of questions, I would like us to discuss how you understand and what you think about existing treatments and the potential cures for sickle cell disease.		
What does good health mean to you?	To investigate the ways in which participants think about health	Do you think anyone can achieve good health? What are some of the ways you try to have good health? What are some of the ways you try to ensure your child has good health?
What are some of the treatment options for sickle cell disease you know about?	To assess participants understanding of treatments for sickle cell disease	How many of these has your child tried? Tell me about some of the ones you've preferred Tell me about some of the ones you didn't try or prefer
Can you tell me about some of your experiences with the treatments your child currently receives?	To investigate participants experiences with the available treatment options.	How do you keep up with these treatments? What are some of the challenges you face?
How would you describe a cure for sickle cell disease?	To investigate participants understanding of and interest in a cure for sickle cell disease	What would it do? What impact do you think it would have on your child's life?

Currently, bone marrow transplantation is the only approved cure for sickle cell disease. Bone marrow makes blood in the body. A donor gives his or her healthy bone marrow to the patient so that he or she can make healthy blood cells.		
Have you heard about bone marrow transplantation?		Is it something that you want for your child? Do you think that it's something that you could have access to? Why?
There are 2 cures for SCD that are currently being developed called gene editing and gene transfer therapy. They make changes to your genes that allow your body to make blood cells that don't sickle. They are still being tested in research studies to see how they work on people with sickle cell disease		
What do you think about treatments that could change your child's genes?	To investigate participants understanding of and interest in a cure for sickle cell disease	What are some concerns you may have? What are some things you would want to know about these kinds of treatments?
Would you let your child have these kinds of treatment?	To investigate participants understanding of and interest in a cure for sickle cell disease	Why or why not?
Part 4- For these final questions I want to go into how people with sickle cell disease are treated in your community.		
Do you feel you are treated differently as the parent of a child with sickle cell disease?	To explore the treatment of parents of individuals with sickle cell disease	In what ways? Positive or negative? What are some of the things people say?

		How does this make you feel? Do you feel that any of them are true?
What do you tell your child about telling others about his or her sickle cell disease status?	To explore the treatment of individuals with sickle cell disease	How do you decide to tell others about your child's sickle cell disease status? Can you tell me about any moments relating to this that have stood out to you?
How do you think sickle cell disease may affect his or her life?	To investigate the participant's perception of sickle cell disease in relation to social and cultural contexts; coping	Do you think it may limit your child in any way? How? Do you think that there are opportunities that sickle cell disease has caused your child to miss out on? How have you tried to help your child overcome these limits? How do you help him or her cope?
How do you cope with the ways that the disease has affected your child?		Do you have a support system (family, friends, religious groups)? If yes, what role do they play in your coping?
How do you imagine your child's future?	Broad question to investigate the participant's outlook with sickle cell disease	
Finally, do you have any questions or anything else you'd like to add? *Thank you again for taking the time to talk with me today*		

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