(Lack of) knowledge of mothers about sickle cell trait and disease: a qualitative study

O (des)conhecimento das mães sobre o trâço e a doença falciforme: um estudo qualitativo

El (des) conocimiento de las madres sobre células falciformes y anemia falciforme: un estudio cualitativo

ABSTRACT
Objective: to analyze the understanding of mothers about sickle cell disease and/or trait of the family from a diagnosed child. Methods: this is a qualitative study, using a semi-structured interview with 23 mothers, at a sickle cell disease outpatient clinic of a public institution, from October to December 2017. Analysis was thematic. Results: all participants had sickle cell trait as well as the parents of their children. Twenty children were diagnosed with sickle cell disease by Heel Prick Test, and three, after hospitalization due to the disease. Most did not know how to report the presence of the trait or disease in relatives other than nuclear. Final considerations: diagnosis cannot be restricted to the result of neonatal screening, requiring that preventive information on sickle cell crises be reinforced. It is recommended to search for other affected relatives to learn about their genetic condition, reflecting on their reproductive decisions. Descriptors: Pediatric Nursing; Neonatal Screening; Heredity; Sickle Cell Trait; Sickle Cell Anemia.

RESUMEN
Objetivo: analizar el entendimiento de madres acerca de la condición del trazón o enfermedad falciforme de su familia a partir de un niño diagnosticado. Métodos: se trata de un estudio cualitativo, mediante entrevista semiestructurada a 23 madres, en el ambulatorio de anemia falciforme de una institución pública, en el periodo de octubre a diciembre de 2017. A analíisis de contenido. Resultados: todos los participantes tenían trazón falciforme, bien como los padres de sus hijos. Veinte niños fueron diagnosticados con enfermedad falciforme, bem como os pais dos seus filhos. Vinte crianças foram diagnosticadas com doença falciforme pelo teste do pezinho, e três, após hospitalização decorrente da doença. A maioria não sabia informar a presença do trazón ou doença em outros membros da família que não a nuclear. Considerações finais: o diagnóstico não pode ficar restrito ao resultado do triagem neonatal, necessitando que as informações preventivas de crises falcêmicas sejam reforçadas. Recomenda-se a busca de outros membros da família afetados para conhecimento da sua condição genética, refletindo sobre suas decisões reprodutivas. Descriptores: Enfermagem Pediátrica; Triagem Neonatal; Hereditariedade; Traço Falciforme; Anemia Falciforme.

RESUMO
INTRODUCTION

The term sickle cell disease refers to the group of hemoglobinopathies related to a genetic and hereditary alteration in hemoglobin S (HbS), which modifies the shape of red blood cells. It may occur in the homozygous form, causing sickle cell anemia (HbSS), but it can also occur from the combination of HbS with other altered types of hemoglobin such as hemoglobin C (HbSC), hemoglobin D (HbSD) and beta-thalassemia (Hb S/β Th)\(^{(1)}\).

Vaso-occlusive phenomena and chronic hemolysis are the main determinants of clinical manifestations and give rise to complications such as chronic anemia, stroke, ocular complications, foot-hand syndrome, sickle cell pain, functional asplenia, growth retardation, atypical secondary sexual characteristics and more frequent and severe infections, which decreases the quality of life of those living with sickle cell disease\(^{(2-4)}\).

Although treatable, sickle cell disease only has the possibility of cure through the transplantation of related allogeneic hematopoietic stem cells, as established by the Ministry of Health, in Ordinance 298 of February 9, 2018\(^{(3)}\). In this sense, it is important that the disease is detected in the family, in order to avoid its heredity through genetic counseling, for the prevention of complications, increased survival, improvement of quality of life and control of disease frequency\(^{(5)}\).

It is estimated that, in Brazil, 4% of the population has sickle cell trait and that 25,000 to 50,000 have compound or double homozygous or heterozygous state\(^{(6)}\). The prevalence of HbAS is higher in the north and northeast, between 6% and 10%, while in the south and southeast, only 2% to 3% of the population is reached\(^{(8)}\), justified by the haplotypes transmitted by the black population coming from Africa, enslaved in the sugarcane crops in the northeast in the extraction of precious metals in Minas Gerais\(^{(7)}\).

It is important to highlight that patients with a gene for HbS and HbA (normal hemoglobin) do not have the disease; however, they have the so-called sickle cell trait (HbAS)\(^{(1-2)}\). They are able to transmit genetically and, if combined with another individual who possesses the trait, can generate a child with the disease in 25% of cases; therefore, it is essential to study the disease heredity.

Recognizing the epidemiological importance of sickle cell disease, the Brazilian National Neonatal Screening Program (PNTN - Programa Nacional de Triagem Neonatal), known as Heel Prick Test, identifies not only newborns affected before the development of any symptoms, but also those who carry sickle cell trait and who can transmit the gene to their children\(^{(9)}\), helping to reduce morbidity and mortality and improve their quality of life. For children and adults who did not take the Heel Prick Test, hemoglobin electrophoresis test\(^{(10)}\) should be offered.

In Brazil, sickle cell disease represents an important public health problem, due to its high prevalence, since it is estimated between 60,000 and 100,000 people living with the disease, whose concentration varies according to the region\(^{(9)}\).

In view of this scenario, services are needed that promote the reception and social support necessary to minimize the difficulties in coping with the disease by welcoming families with responsibility and resolution, promoting the formation of bonds and minimizing the implications generated by the disease\(^{(10)}\). Moreover, the knowledge obtained by the family about its genetic transmission is considered fundamental as individuals should be informed about their health to better analyze their reproductive decisions\(^{(10)}\).

Genetic counseling is an important tool in the field of hereditary diseases, as it encourages implementing preventive care practices and guidance to parents in relation to newborns. It addresses educational and reproductive aspects, which are essential for improving the life of patients with sickle cell disease and also allow pedagogical action on the family’s genetic condition as well as the risk of recurrence in future pregnancies through family guidance or genetic counseling\(^{(11)}\).

It is noteworthy that, for adults living with sickle cell disease or trait, it is essential to perform preconception genetic counseling, because there is a chance that children will inherit sickle cell anemia and thalassemia\(^{(12)}\).

The study on reproductive decisions from the perspective of women caregivers of children with sickle cell disease inserted in a Federal District’s neonatal screening program, pointed out that, even with the possibility of having other children with sickle cell disease or trait, they wished to have more children. However, women who do not want to have more children and women whose reproductive project is still being developed were also found, based on information received in the screening program\(^{(13)}\).

This study concludes that women’s reproductive choices are not built solely on biomedical information about the disease, but on the experience of caring for children and themselves. On the other hand, some reports suggest that some women maintain a discursive strategy based on the medicalization of reproductive choices, stating that they did not have any more children due to having undergone tubal ligation or that the partner had a vasectomy. They also affirm the scarcity of publications addressing the reproductive risk associated with sickle cell disease, despite the ethical and social epidemiological centrality of the disease\(^{(13)}\).

Sickle cell trait is a recessive genetic condition, in which knowledge is essential for people to properly decide on their reproductive decisions\(^{(14)}\).

Two important factors stand out, early diagnosis and genetic counseling. In this context, it is essential to approach the knowledge learned by the families of children with trait or sickle cell disease about their condition. Then, the following guiding questions arose: which relatives have the trait or have sickle cell disease? What is the relative’s knowledge of the trait or sickle cell disease in the family?

The object of the study is the (lack of) knowledge of mothers about sickle cell disease or trait in the family.

OBJECTIVE

To analyze the understanding of mothers about sickle cell disease and/or trait of the family from a diagnosed child.

METHODS

Ethical aspects

As it is a research involving human beings, the present study was assessed by the Institutional Review Board (IRB) of the proposing
机构，以保持参与者匿名性，它们被识别为母亲的字母M，按照访谈序列号。

**Type of study**

这是一项描述性研究，具有定性方法，因为它试图在一个意义上工作，即意义、动机、愿望和态度，以及回答特定问题，基于家庭的现实和个人性。

为了保持研究方法论的严谨性，根据家庭的现实和个别性，它使用了报告定性研究的整合化标准。

**Study setting**

研究是在血液科门诊处进行的，位于里约热内卢的一个市镇。

它是一个为诊断和治疗儿童而设立的参考机构。

**Study participants**

该研究包括了23名母亲，她们是陪伴患有CDA或CDS的儿童到医疗或牙科咨询，并自愿想参与这项研究的。

在医院期间，通常是由母亲来照顾患病儿童。

参与者是他们孩子的CDA或CDS的诊断，以及在研究中被接受和治疗的。

在血液科门诊处进行的访谈中，母亲和孩子被排除，考虑到他们可能没有父母遗传条件。

**Data collection and organization**

收集数据和组织

对于数据收集，使用了个人访谈和半结构化访谈脚本，其中问题为：哪些亲属有CDA或CDS？你了解有关你孩子的CDA或CDS吗？你如何了解有关你孩子的CDA或CDS？

数据收集始于通过预约参与者的门诊咨询日，此前预约通过预约日历。在该日，当潜在参与者被识别时，他们被通知了研究的目标。

访谈的开始是邀请到一个之前被识别的房间，房间是空的。

在预约当天，从8点到10点，访谈是按预约时间进行的；与医生的咨询从10点开始，因此，8点到10点，访谈是与医生的。

在进行访谈时，检查表和确认文件由参与者签署，一份副本。

**Data analysis**

数据分析

在转录后，数据分析是主题的，根据预分析、材料探索、治疗和解释的获得结果阶段(17-18)，构建了以下分析类别，即“识别CDA或CDS家族：(缺乏)知识在面对遗传性”和“拇指刺穿测试结果：诊断影响和处理情况”，这将呈现在下面。

**RESULTS**

识别CDA或CDS家族：(缺乏)知识在面对遗传性

这个分析类别呈现了有CDA或CDS的亲属，根据参与者的资料。其中23名母亲受访者中，所有受访者都发现有CDA。

根据一些报告，有缺乏遗传知识，CDA或CDS的亲属。

**Participants acknowledge that the parents of the child accompanied by the hematology outpatient clinic carry sickle cell trait and have at least one child with sickle cell disease.**
the child’s grandfather, uncles and cousins have sickle cell trait or even beta-thalassemia, on the other hand, other mothers report not knowing this information. In this context, it is noteworthy that one participant only discovered that other relatives had sickle cell disease after the niece’s death (child’s cousin).

It appears that, of the 23 interviewees, fifteen had only one child, all with sickle cell disease. Four of the participants had two children, one child of one of the participants had sickle cell trait, while the other three mothers had the other child without the trait and without the disease. Another four participants had three children, and it is noteworthy that, of these, one mother reported having a child with sickle cell disease and the other without the trait or disease, while the other three mothers had at least one child with sickle cell trait, one of them had, in addition to a son with the trait, another son with beta-thalassemia. It can be inferred from this result that the greater the number of children, the greater the probability of generating new children with sickle cell trait or disease.

The Heel Prick Test result: impact of diagnosis and coping with the situation

This analysis category presents the knowledge of sickle cell disease through the Brazilian Neonatal Screening program/Heel Prick Test; the impact of the moment of diagnosis and the search for information; the therapeutic itinerary and feelings related to complications; sequel of sickle cell disease as well as their care and medications used.

Most participants report that they had knowledge of sickle cell disease in their child through the Brazilian Neonatal Screening program/Heel Prick Test, as highlighted by some statements:

In her Heel Prick Test, it was accused that she had sickle cell anemia [...] then I got to know more deeply [...]. (M2)

I found out as soon as she was born, then I did the Heel Prick Test, then I had this anemia [...] I didn't know about the existence of this disease. (M7)

Through the Heel Prick Test, as soon as she was born [...] because my other son has the trait and then I was more or less aware of things, then I knew that when I had another child I could have come with anemia [...]. (M14)

[...] Heel Prick Test [...] in adolescence, I took the test that had sickle cell trait, then, after I got pregnant, I asked my husband to do the test too, then we went to the hematologist [...] then we found out that he [child] had anemia [...] then he did the Heel Prick Test at 5 days old [...]. (M22)

I found out through the Heel Prick Test [...] I didn’t know what sickle cell anemia was, I found out after her [...]. (M23)

It is noteworthy that M22 asked the child’s father to take the test only during pregnancy, even though he knew he had sickle cell trait since adolescence. It is noteworthy that the majority of participants report being informed about sickle cell trait or disease, when receiving the test result. However, it appears that they were not prepared to effectively understand the information regarding sickle cell trait or disease inheritance.

On the other hand, this information can determine the decision to know before planning a child about genetic issues, as highlighted in M21’s speech:

The only thing that happened was that my brother learned that my niece, who is now 20 years old, could not have a child [...], that before getting married it would be ideal, ideal, for the groom to take the test to find out if he would have the trait. (M21)

Three mothers reported that the child took the Heel Prick Test, but they only discovered their child’s disease when they had their first flare-up, according to the following reports:

In his Heel Prick Test, it was already written that he had sickle cell anemia, he already had very low hemoglobin and hematocrit rates, so she asked to take him to a hospital [...] he was 1 and a half years old when he found out [...]. (M5)

C15 was sick at 3 months and I took him to several hospitals, and the Heel Prick Test was not ready [...], so, sickle cell anemia was discovered here at the hospital [...]. (M15)

Her Heel Prick Test was lost, there was no result, then I was given the phone number to call at the time [...] called several times. Then, when she was 2 years old, it was her first crisis, she went down wanting to pass out, then she went to another hospital [...] then they found out. Her father and I had to collect blood to confirm the trait [...]. (M21)

It is assumed that there is no appreciation of this information by health professionals when they do not talk to families, in order to remove doubts and know about the understanding of the guidelines, not making efforts to show the importance of this diagnosis. Consequently, families only seek help when the child with the disease goes into crisis.

On the other hand, some participants reported exactly that, when the Heel Prick Test result was positive, they were contacted by the reference hospital to repeat it:

[...] when they called from the health department who had diagnosed a disease, at the time, they did not say what it was [...] they sent us to the hospital, it was well explained [...]. (M3)

I was suspected of sickle cell anemia [...] they called me saying that there was a change in her Heel Prick Test, then I went there, they called from the hospital [...]. (M8)

[...] the Heel Prick Test [...] it took a little while to arrive and when it arrived it was inconclusive, they explained to me that it could be sickle cell trait [...] and indicated that I go to the hospital, I went with faith, but unfortunately it was positive [...] for the disease [...]. (M18)

It is a fact that, when relatives are well informed and with no outstanding doubts, it becomes a fundamental element for children’s health, minimizing the sequels and crises of sickle cell disease throughout the life of this individual.

As for the itinerary of children with sickle cell disease, practically all participants reported having obtained the first statement from the reference hospital, where the Heel Prick Test was collected; subsequently, they were referred to the study scenario for follow-up, according to some statements:
The maternity unit called saying that I had an appointment at the hospital, so I went there, and sent it from there, you know? Then treatment began. (M6)

As soon as the result came..., they were referred to APAE [Association of Parents and Friends of Disabled Individuals] from where she lived... then I arrived here she was 7 months old, I came to seek care... and from here they continued to treat her today... (M17)

Participants reported an impact at the child's diagnosis time, reporting feelings such as fear, insecurity, denial and sadness, according to some statements:

So, when we found out, there was desperation, desperation, first it's something we don't know [...]. Because, when the doctor explains to us, he said, "The child cannot go to the beach, he cannot play soccer, he cannot practice any fight, no sport, he must always be hydrated, he cannot take a cold shower," and we start to get desperate, so he cannot do anything? We have to go testing through limits. (M8)

When he said that my daughter had this disease, I didn't believe it, the test came wrong, because I didn't know I had the trait nor did her father had the trait, so when they took the exam there at the hospital, they repeated again, here they repeated it again it really has, we now realized, period. Wow, this disease is the end of the world [...]. (M13)

The way I heard, it was horrible [...]. It was a social worker who spoke to me, she just said, "Mother, she has this disease, it is a blood disease, she will treat it for the rest of her life, her son needs to take Benzetacil, folic acid, which SUS only offers only Benzetacil, that you cannot let your child have this or that, he cannot have fever, rubella, chicken pox, measles, he cannot be a child like any other" [...]. (M16)

It was sad to know that he had sickle cell anemia, especially knowing that this disease has no cure, so it is complicated to know [...]. we fight for it, for her survival [...]. (M23)

In the statements described, it appears that the information was provided by the doctor and the social worker; however, this result leads us to affirm the need for the participation of a multidisciplinary team, with greater participation of a nurse, and that this information be passed on gradually, because these are difficult to be processed. The information that the professional considers important, often at the child's diagnosis time, is not the demand for information that the relative seeks, including, because, if the child is not in the sickle cell crisis, there are no major changes in care, as the following report:

In my life, there was no impact, nor did the others have the trait, the impact was great in my son's question, we didn't change anything, but for him it was a big change [...]. (M3)

In view of sickle-cell crises, it appears that relatives are concerned with what their children suffer or may suffer in the future:

[...] it is very complicated to see your child suffering, dying of pain or always having to keep an eye on him, that he may have a crisis [...]. we suffer, we have to be around the clock, 24 hours a day attention on him. (M20)

I know he may have a bone pain crisis, need a transfusion, so, like this, we are worried that he will suffer [...]. (M22)

Some precautions were reported by the participants in order to avoid sickle cell crisis, which are to avoid places with very low temperatures, not to allow the use of a swimming pool, to identify jaundice and splenomegaly, and to be aware that, if children present any other symptom, they should be referred to the hospital:

I can't go out with her to colder places anymore, I can't go to the pool [...]. (M2)

As in this disease, hemoglobin does not have a normal shape, circulation is limited, so, with hydration, it helps a lot in the issue of circulation. (M4)

[...] anything, you have to run to the hospital [...]. if so, we will discover the symptoms, such, these things, hand or something that is swollen and [...] pale, fever, if you had a fever you have to come here [...]. (M6)

[...] very careful, there are many things that appear [...] you cannot be drinking ice cream, because the flu can become very easy pneumonia, in this case [...]. (M11)

[...] I know how to palpate the spleen to see if the spleen is enlarging; when her hemoglobin is falling, her eye is yellow, her skin is a little yellow [...]. But when she is really sick [...] I run [...]. (M12)

Participants demonstrate having obtained information about the signs and symptoms of sickle cell crisis or clinical situations that can lead children to an emergency or hospitalization condition, demonstrating awareness of children's treatment in order to avoid serious complications of the disease.

DISCUSSION

Currently, the Heel Prick Test, carried out through PNTN, is one of the main ways to diagnose sickle cell disease and other hemoglobinopathies that, when identified early, improve children's survival conditions. The Brazilian Unified Health System (SUS – Sistema Único de Saúde) guarantees specialized care to all screened children through 31 neonatal screening services in the country, distributed among all Brazilian states. The Heel Prick Test can be performed at 21,446 collection points, distributed in Primary Care, Hospitals and Maternity network(19) and it is the child's right(20).

The path taken until reaching this scenario was the creation of the Hemoglobinopathies Committee by the Ministry of Health in 1992, responsible for the first measures to publicize and standardize the treatment of these diseases in our country. On May 10, 1996, the working group created by the Ministry of Health Ordinance 951/96, coordinated by the Brazilian National Health Regulatory Agency (ANVISA - Agência Nacional de Vigilância Sanitária), implemented, in August, the Sickle Cell Anemia Program (SCAP). Sickle cell anemia once again gained political support in the Ministry of Health in 2001 by PNTN(21).

In 2004, the Coordination of the Brazilian National Policy for Blood and Blood Products (Política Nacional do Sangue e Hemoderivados) was created, a sector in charge of outlining a policy.
for the care of sickle cell disease and other hemoglobinopathies in SUS, as recommended by Ordinance MO 1.391/05, aiming at promoting a change in the natural history of sickle cell disease in Brazil, reducing the rate of morbidity and mortality, promoting longevity with quality of life for people with this disease, guiding those with sickle cell trait and informing the general population.

Sickle cell trait is associated with the risk of birth of future children with the disease; therefore, knowing the epidemiological profile with regard to hemoglobinopathies in the Brazilian population is important to establish the health care network and provide genetic counseling to patients and their families.

Furthermore, it favors behaviors that reduce the risk and vulnerability to changes in child development, preventing various complications and sequelae.

With information on the disease heredity, it is possible to make a reproductive planning in order to avoid the birth of children with the disease as well as allowing the free choice of parents in the decision to have or not a child. The importance of a multidisciplinary health team that can discuss together with the demands of these families, solve doubts, provide guidance regarding preventive care for sickle cell crises, or even refer them to referral hospitals so that they can be assisted properly.

There is still a need for an expansion of specialized institutions in regions with little access or rural areas, in order to serve children in a location close to their homes, and to improve the articulation of hospitals with primary or rehabilitation care, so that it can be informed about the test result and be monitored frequently in a non-fragmented way.

Another challenge is the therapeutic itinerary, which, due to families' low financial conditions, can make it difficult to access the diagnosis on their return and the consequent follow-up of treatment, which can bring complications.

In the adult population living with sickle cell disease or trait, the importance of preconception genetic counseling is emphasized, because of the chances of children inheriting sickle cell trait, sickle cell disease and thalassemia.

In this context, it is necessary to highlight that many of the people born before 2001 discovered late their condition as carriers of sickle cell trait; in other words, this occurred only when their child was diagnosed with a disease or trait, which made it impossible for these people to access conscious family planning choices.

The study sought to reveal knowledge about sickle cell disease in terms of its causes, mode of transmission and factors that increase or decrease the chance of being a carrier or having the disease. It also evidenced that the majority of participants knew that it was a hereditary disease, but only 31.3% agreed that it was useful to know if they are carriers, if they have a sick partner or if they have a sickle cell trait. On the other hand, more than half did not consider this information.

The aforementioned study demonstrated the relationship between lack of knowledge and bad attitudes or practices towards sickle cell disease by revealing that half of the studied population demonstrated little knowledge about sickle cell disease and only a quarter demonstrated good attitudes and practices.

There is also a concern of mothers regarding stigmatization of the disease due to their Afro-descendant origin. Another fact that deserves to be highlighted is the stigmatization of the disease, since the racial association caused by the disease itself affects the person's well-being and quality of life, one of the possible impediments to the dissemination of information about heredity in the family environment.

Assistance in the genetic counseling service, in turn, is relevant not only in terms of future reproductive decisions, but also in terms of identifying people who need help to adjust to their relative's illness or their own illness, with availability of this service that starts in primary care to a specialized reference service.

Considering sickle cell trait and disease a condition of heredity, this type of care becomes relevant. However, it should be noted that, in practice, genetic counseling services are restricted to specialized institutions, contributing to the difficulty of accessing this service and to the length of the Brazilian National Policy for Comprehensive Care in Clinical Genetics (Política Nacional de Atenção Integral em Genética Clínica), created since 2009.

It is necessary to highlight that, in genetic counseling, the information must be accurate and objective about the implications, advantages and disadvantages for the possible parents and their probable children, in the face of the family's decision to have them or not. In any case, genetic counseling must be non-directive and impartial, and parents' decisions must be supported, regardless of reproductive choice.

Families who experience sickle cell disease struggle daily to adapt to the condition of having a member affected by a chronic disease, as their routines are modified to care for sick children. The attention that parents pay to the care of their children allows them to deal with the reality of an injury and is part of family life, which makes them important references in the care and survival with quality of their children. In this context, families tend to outline a therapeutic itinerary, which is considered as an itinerary of individuals by health services, and the sequence of decisions and events, building a determined trajectory with the objective of treating the disease or preventing their crises.

Relatives of children with sickle cell disease must be informed about the disease, its repercussions and the care they will demand throughout their lives, in order to avoid complications and early deaths. It can be inferred that the disease causes changes in the family's routine, bringing greater responsibility in care and treatment of children with sickle cell disease. The complications and sequelae of the disease are the main concerns of those responsible.

Among the complications of sickle cell disease, vasocclusive crisis, priapism, ischemic bone necrosis, kidney disease, acute chest syndrome, stroke, severe anemia and retinopathy can be mentioned, which can result in reduced life expectancy for people with the disease compared to the general population.

In the present study, participants cited the use of antibiotics as one of the measures to prevent complications. According to the Ministry of Health, several actions can contribute significantly to reduce mortality among the population living with sickle cell disease, including the control of infections through immunizations and the prophylactic use of antibiotics in the first years of life. Added to these actions is the ability of relatives to identify the important signs and symptoms presented by children and to seek immediate assistance, as in the case of splenic kidnapping, diagnosis and appropriate treatment in several other emergency situations.
In view of family changes and the fragile health status of children with sickle cell disease, monitoring and support offered by health professionals is recommended, as these children need surveillance in the community that can be developed by primary care services.

Study limitations

The limitation is in the fact that it was carried out with some relatives of children with sickle cell disease followed up on an outpatient basis in a single pediatric institution, that, despite being a reference for sickle cell disease, the results cannot be generalized, representing only the outcome of a group of participants.

Contributions for health

It was found in mothers’ reports lack of information about the heredity of sickle cell disease, despite the existence of public policies both for early diagnosis, through neonatal screening, and for its treatment. It is hoped that this study will contribute to the visibility of sickle cell disease and the importance of genetic counseling so that people can find out if they have sickle cell trait, assisting them in their reproductive choices, as it is an inherited genetic disease.

FINAL CONSIDERATIONS

The 23 participants were mothers and found, both fathers, to have sickle cell trait in view of diagnosis of their children with the disease through neonatal screening. Fourteen of these mothers had only one child with sickle cell disease and the other nine mothers had one or two children, resulting in at least one child with sickle cell trait, sickle cell disease or beta-thalassemia as well as children who did not even have the trait nor the disease.

It appears that, despite being a genetically transmitted disease, study participants (do not) know their inheritance in other relatives. Few were those who reported having knowledge of sickle cell trait in grandparents, uncles or cousins. Due to the lack of knowledge about their genetic condition and that of their families, people may be deprived of making conscious choices regarding their sexual and reproductive health, due to the lack of access to genetic counseling services.

On the other hand, they were able to recognize the predictive signs of a sickle cell crisis and the main signs and symptoms in their children who have the disease. Still, they mention knowing when to take them to the hospital or health units, in the case of an emergency, highlighting some care related to children’s body temperature, hydration and physical activities as well as the use of prophylactic medications.

Considering the results, it is necessary to rethink and discuss public policies focused on the theme regarding the educational issue and/or further clarification to the population. Greater investment should also be considered for the qualification of health professionals and the expansion of genetic counseling services in public and private institutions.

Diagnostic information about sickle cell trait or disease cannot be restricted to the result of neonatal screening, requiring the information to be extended to child care, preventing sickle cell crises and other diseases. For these families, it is also recommended to search for other affected members to learn about their genetic condition, in order to give autonomy over the reproductive decisions of future generations.

As it is a disease linked to heredity and African ethnicity, as well as being associated with social and economic conditions, it is a great challenge for citizens and health professionals to implement public policies that can address issues so complex that go beyond the conditions of health. It is also necessary to improve genetic counseling strategies and greater investment that can address issues that go beyond the collection of material and the result of the Heel Prick Test in the national program.

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