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

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

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

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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.

RESUMO

Objetivo: analisar o entendimento de mães acerca da condição do traço e/ou doença falciforme da família a partir da criança diagnosticada. **Métodos:** trata-se de um estudo qualitativo, sendo utilizada entrevista semiestruturada, com 23 mães, no ambulatório de doença falciforme de uma instituição pública, no período de outubro a dezembro de 2017. A análise foi temática. **Resultados:** todos os participantes tinham traço 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 traço ou doença em outros membros da família que não a nuclear. **Considerações finais:** o diagnóstico não pode ficar restrito ao resultado da 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.
Descritores: Enfermagem Pediátrica; Triagem Neonatal; Hereditariedade; Traço Falciforme; Anemia Falciforme.

RESUMEN

Objetivo: analizar el entendimiento de las madres sobre células y/o anemia falciformes de la familia del 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, de octubre a diciembre de 2017. El análisis fue temático. **Resultados:** todos los participantes tenían células falciformes, así como los padres de sus hijos. Veinte niños fueron diagnosticados con anemia de células falciformes mediante la prueba de punción del talón y tres, después de la hospitalización debido a la enfermedad. La mayoría no sabía cómo informar la presencia de células o anemia en miembros de la familia distintos del nuclear. **Consideraciones finales:** el diagnóstico no puede restringirse al resultado del cribado neonatal, requiriendo que se refuerce la información preventiva sobre las crisis drepanocíticas. Se recomienda buscar a otros familiares afectados para conocer su condición genética, reflexionando sobre sus decisiones reproductivas.
Descriptores: Enfermería Pediátrica; Tamizaje Neonatal; Herencia; Células Falciformes; Anemia Falciforme.

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 sare 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, atin 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[3]. 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[6], 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[8], 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[3] 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[8].

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

institution, approved. To maintain participants' anonymity, they were identified with the letter M of mother, followed by the sequence number of the interviews.

Type of study

This is a descriptive study with a qualitative approach, because it seeks to work with a universe of meanings, motives, aspirations, beliefs, values and attitudes as well as to answer particular questions based on families' reality and individuality.

To maintain the methodological rigor of the study, the Consolidated criteria for reporting qualitative research (COREQ)[15] was used as a support tool.

Study setting

The research was carried out at a hematology outpatient clinic of a public teaching, research and care institution specialized in pediatrics, located in a municipality of Rio de Janeiro.

It is a reference institution for diagnosis and care of children, who are incomplete with incomplete 29 days to 13 years with hematological problems, including sickle cell disease.

Study participants

The study included 23 mothers who were accompanying the child with sickle cell disease to medical or dental consultation and who voluntarily wished to participate in the study after invitation. During hospitalization, it is usually mothers who take care of sick children.

Participants who were aware of their children's sickle cell disease diagnosis and who were registered, being followed up to the hematology outpatient clinic in the study setting were included. Adoptive mothers were excluded, considering that they might not have knowledge of parents' genetic condition for sickle cell trait or disease.

Data collection and organization

For data collection, individual interview and a semi-structured interview script were used, whose questions were: which relatives have sickle cell trait or disease besides your child? How did you learn about your son's sickle cell disease and what do you know about this disease or sickle cell trait in your relatives?

Data collection was initiated by contacting participants at the outpatient consultation day, previously identified through scheduling book. On the day when the prospective participants were identified, they were informed about the objectives of the study and invited to be volunteers. Once the invitation was accepted, they received the Informed Consent Form (ICF), which was read with the researcher. Once all doubts were clarified, the document was signed by participants, with a copy of it.

Subsequently, they were referred to a room previously reserved for the interview, in the outpatient sector, a room that is free from the transit of people. It is noteworthy that the relatives who accompanied their children at the clinic arrived at 8 a.m. to weigh and measure them; consultations with the doctor started at 10 a.m.; therefore, between 8 a.m. and 10 a.m., interviews were conducted. When this was not possible, the participants were interviewed right after the medical consultation.

Collection was always performed on Thursdays, the hematology outpatient clinic day, from October to December 2017. It is noteworthy that the interview script was validated in the first interview, and there was no need to adjust the questions. The interviews were recorded with the aid of a portable recorder and lasted an average of 10 minutes.

The interviews ceased when there was theoretical saturation, which, in qualitative research, is a fundamental for interrupting the recruitment of participants and defining the sample size[16].

Data analysis

After transcribing the statements, data analysis was thematic, following the pre-analysis, material exploration, treatment and interpretation of the obtained results stages[17-18], constructing the following categories of analysis, "Identification of sickle cell disease or trait in the family: (Lack of) knowledge in the face of heredity" and "The Heel Prick Test result: impact of diagnosis and coping with the situation", which will be presented below.

RESULTS

Identification of sickle cell disease or trait in the family: (Lack of) knowledge in the face of heredity

This category of analysis presents relatives who have sickle cell trait and/or disease according to participants' information. Of the 23 mothers interviewed, all of them found to have sickle cell trait, as well as the parents of their children, through the children's diagnosis.

As highlighted by some reports, there is a lack of genetic knowledge about the disease or sickle cell trait in other relatives, in addition to the nuclear one:

> *Me, my mother, the father and the grandfather, the father of my husband* [...] *we didn't even know we had the trait* [...]. (M1)

> *We only discovered the trait after she performed the Heel Prick Test, so I already commented with several relatives, but nobody went to find out, so I have no idea if it already existed in the family. I have another son who has none of this.* (M11)

> *There was no family screening, only the screening was done between me and the father* [...] *we don't know about other people in the family who have the trait or the disease.* (M16)

> *I have and her father has the trait* [...] *a niece who has passed away had sickle cell anemia, but the mother only found out later, at the time she did not do the Heel Prick test, when she passed away, I think she was 3 years old.* (M19)

> *In my family, I, my older brother and a niece have the trait, my other brother has the thalassemia trait and his son too.* (M20)

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. If, on the one hand, some mothers report knowing that other relatives such as

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 children 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* [...] *I 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[22].

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[12]. Furthermore, it favors behaviors that reduce the risk and vulnerability to changes in child development, preventing various complications and sequels[23].

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[24].

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[12].

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[25].

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[24]. 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[26-27].

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[28]. 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[29].

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[29]. In any case, genetic counseling must be non-directive and impartial, and parents' decisions must be supported, regardless of reproductive choice[30].

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[31]. 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[32].

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[32]. 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 sequels 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[3].

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[3].

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.

REFERENCES

1. Arduini GAO, Rodrigues LP, Marqui ABT. Mortality by sickle cell disease in Brazil. Rev Bras Hematol Hemoter. 2017;39(1):52-6. https://doi.org/10.1016/j.bjhh.2016.09.008
2. Ware R, Montalembert M, Tshilolo L, Abboud MR. Sickle cell disease. Lancet. 2017;390(10091):311-23. https://doi.org/10.1016/S0140-6736(17)30193-9
3. Ministério as Saúde (BR). Comissão Nacional de Incorporação de Tecnologias no SUS. Protocolo Clínico e Diretrizes Terapêuticas Doença Falciforme. Brasília: Ministério da Saúde, 2018.
4. Oliveira P, Santos K, Silva F, Dias A, Silveira E, Guimarães, E. Evaluation and intervention in the family of adolescents with sickle cell disease. Ev Enferm UFPE. 2017;11(4):1552-64. https://doi.org/10.5205/1981-8963-v11i4a15223p1552-1564-2017
5. Souza I, Araújo E. Doença falciforme e triagem neonatal: um debate necessário. Rev Saúde Col UEFS. 2016;5(1):59-61. https://doi.org/10.13102/rscdauefs.v5i1.1011
6. Sarat CNF, Ferraz MB, Ferreira JM, Corrêa FRAC, Souza AS, Cardoso AIQ, et al. Prevalence of sickle cell disease in adults with delayed diagnosis. Acta Paul Enferm. 2019;32(2):202-9. https://doi.org/10.1590/1982-0194201900028
7. Calvo-González E. On slaves and genes: "origins" and "processes" in genetic studies of the Brazilian population. Hist Cienc Saude-Manguinhos. 2014;21(4):1113-29. https://doi.org/10.1590/S0104-59702014000400003
8. Leite DCF, Cipolotti R, Gurgel RQ, Martins FPRS, Lopes GD. Spatial distribution of newborns with sickle cell trait in Sergipe, Brazil. Rev Paul Pediatr. 2020;38:e2018229. https://doi.org/10.1590/1984-0462/2020/38/2018229

9. Ministério da Saúde (BR). Tratamento para Doença Falciforme está disponível no SUS [Internet]. 2019 [cited 2020 Jul 10]. Available from: http://www.blog.saude.gov.br/l1zfx9

10. Machado A, Sousa M, Silva M, Coutinho S, Reichert A, Collet N. Difficulties in effecting hospital reception at admission of children with chronic disease. Rev Enferm UERJ. 2015;23(4):556-61. https://doi.org/10.12957/reuerj.2015.5960

11. Ministério da Saúde (BR). Secretaria de Atenção a Saúde. Departamento de Atenção Especializada e Temática. Triagem neonatal biológica: manual técnico. Brasília: Ministério da Saúde, 2016.

12. Rosenfeld LG, Bacal NS, Cuder MAM, Silva AG, Machado ÍE, Pereira CA, et al. Prevalence of hemoglobinopathies in the Brazilian adult population: National Health Survey 2014-2015. Rev bras Epidemiol. 2019;22(Suppl2):e190007. https://doi.org/10.1590/1980-549720190007.supl.2

13. Guedes C. Reproductive decisions and newborn screening: the perspective of female caregivers of children with sickle cell disease. Ciênc Saúde Coletiva. 2012;17(9):2367-76. https://doi.org/10.1590/S1413-81232012000900017

14. Mayo-Gamble TL, Barnes PA, Erves JC, Middlestadt SE, Lin HC. 'It means everyone should know their status': exploring lay conceptions of sickle cell trait and sickle cell trait screening among African Americans within middle reproductive age. Ethn Health. 2018;23(7):813-29. https://doi.org/10.1080/13557858.2017.1295135

15. Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ):a 32-item checklist for interviews and focus groups. Int J Qual Health Care. 2007;19(6):349-57. https://doi.org/http://intqhc. oxfordjournals.org/content/19/6/349.long

16. Nascimento LCN, Souza TV, Oliveira ICS, Moraes JRMM, Aguiar RCB, Silva LF. Theoretical saturation in qualitative research: an experience report in interview with schoolchildren. Rev Bras Enferm. 2018;71(1):228-233. https://doi.org/10.1590/0034-7167-2016-0616

17. Minayo MCS, Deslandes SF, Gomes R, (Orgs.). Pesquisa Social: Teoria, método e criatividade. Petrópolis: Vozes; 2016. 96p.

18. Minayo MCS. O desafio do conhecimento: pesquisa qualitativa em saúde. São Paulo: Hucitec; 2014. 215 p.

19. Ministério da Saúde (BR). Exames de triagem neonatal: Teste do Pezinho [Internet]. 2017 [cited 2020 Jul 20]. Available from: https://www.saude.gov.br/artigos/823-assuntos/saude-para-voce/40773-exames-da-triagem-neonatal

20. Senado Federal (BR). Coordenação de Edições Técnicas. Estatuto da criança e do adolescente [Internet]. Brasília; 2017 [cited 2020 Mar 20]. Available from: https://www2.senado.leg.br/bdsf/bitstream/handle/id/534718/eca_1ed.pdf

21. Rodrigues DOW, Ferreira MCB, Campos EMS, Pereira PM, Oliveira CM, Teixeira MTB. History of newborn screening of sickle cell disease in Brazil - the Minas Gerais state chapter. Rev Med Minas Gerais [Internet]. 2012 [cited 2020 Jun 10];22(1):66-72. Available from: http://www.rmmg.org/artigo/detalhes/128

22. Mendes CA, Guigen AP, Anastácio-Pessan FL, Dutka JCR, Lamônica DAC. Knowledge of parents regarding newborn screening test, after accessing the website "Babies' Portal" - Heel prick test. Rev CEFAC. 2017;19(4):475-83. https://doi.org/10.1590/1982-021620171949616

23. Gesteira ECR, Szylit R, Santos MR, Farialchikawa CR, Oliveira PP, Silveira EAA. Family management of children who experience sickle cell disease: a qualitative study. Rev Bras Enferm. 2020;73(suppl 4):e20190521. https://doi.org/10.1590/0034-7167-2019-0521

24. Daak AA, Elsamani E, Ali EH, Mohamed EH, Fatma A, Abdel-Rahman ME, Elderdery AY, Talbot O, Kraft P, Ghebremeskel K, Elbashir MI, Fawzi W. Sickle cell disease in western Sudan: genetic epidemiology and predictors of knowledge attitude and practices. Trop Med Int Health. 2016;21(5):642-653. https://doi.org/10.1111/tmi.12689

25. Xavier EC, Rocha KB. Subjetividade e interseccionalidade: experiências de adoecimento de mulheres negras com doença falciforme. Av Psicol Latinoam. 2017;35(2):267-82. https://doi.org/10.12804/revistas.urosario.edu.co/apl/a.3804

26. Wesley KM, Zhao M, Carroll Y, Porter JS. Caregiver perspectives of stigma associated with sickle cell disease in adolescents. J Pediatr Nurs. 2016;31(1):55-63. https://doi.org/10.1016/j.pedn.2015.09.011

27. Mendes Á, Paneque M, Sousa L, Clarke A, Sequeiros J. How communication of genetic information within the family is addressed in genetic counselling: a systematic review of research evidence. Eur J Hum Genet. 2016;24(3):315-325. https://doi.org/10.1038/ejhg.2015.174

28. Cebulski JN da S, Couto LL do. What to tell the family? Predisposition for hereditary childhood cancer. RSD. 2020;9(11):e56791110056. https://doi.org/10.33448 / rsd-v9i11.10056

29. Pergament E, Pergament D. Reproductive decisions after fetal genetic counseling. Best Pract Res Clin Obstet Gynaecol. 2012;26(5):517-29. https://doi.org/10.1016/j.bpobgyn.2012.04.001

30. Silva AH, Bellato R, Araújo LF. Cotidiano da família que experiência a condição crônica por anemia falciforme. Rev Eletr Enf. 2013;15(2):437-46. https://doi.org/10.5216/ree.v15i2.17687

31. Aquino RCA, Rodrigues M. Acesso e itinerário terapêutico dos pacientes com câncer: principais dificuldades enfrentadas para este percurso. Rev Saúde Com [Internet]. 2016 [cited 10 Aug 2020];12(1):488-96. Available from: http://periodicos2.uesb.br/index.php/rsc/article/view/400

32. Vieira FS, Moreira TMM, Mota CS, Oliveira RS, Gomes ILV. Creation and validation of a health guidance booklet for family members of children with sickle cell disease. Esc Anna Nery. 2019:23(1):e20180231. https://doi.org/10.1590/2177-9465-ean-2018-0231