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ORIGINAL ARTICLE

SOCIODEMOGRAPHIC CHARACTERISTICS AND EPIDEMIOLOGICAL OF CHILDREN WITH SICKLE CELL DISEASE

CARACTERÍSTICAS SOCIODEMOGRÁFICAS E EPIDEMIOLÓGICAS DE CRIANÇAS COM ANEMIA FALCIFORME

CARACTERÍSTICAS SOCIODEMOGRÁFICAS Y EPIDEMIOLÓGICAS DE NIÑOS COM ANEMIA FALCIFORME

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ABSTRACT

Objective*:** to characterize children with sickle cell anemia concerning socioeconomic and epidemiological aspects. ***Method*:** quantitative epidemiological study, conducted through a survey of the records of a public hospital in Teresina/PI, Northeastern of Brazil. The subjects were children aged 0 to 10 years old hospitalized for sickle cell anemia, from December 2010 to March 2011, when the data collections were performed after approval of the project by the Research Ethics Committee, CAAE 0315.0.045.000-10. ***Results*:** data analysis revealed balance in carrying out the diagnostic in Piauí, although the State did not rely on the identification of this disease by Newborn Screening Test, and is still in Phase I, which only covers screening for Congenital Hypothyroidism and Phenylketonuria. ***Conclusion: there are still difficulties in the diagnosis of the disease, mainly because much of the affected reside within the state. ***Descriptors*:** Anemia Sickle Cell; Child Care; Nursing.

RESUMO

***Objetivo*:** caracterizar crianças com anemia falciforme quanto aos aspectos socioeconômicos e epidemiológicos. ***Método*:** estudo de natureza quantitativa epidemiológica, realizado por meio de levantamento em prontuários de um Hospital Público em Teresina/PI, Nordeste do Brasil. Os sujeitos foram crianças de 0 a 10 anos internadas por anemia falciforme, de dezembro de 2010 a março de 2011, quando foram realizadas as coletas de dados, após aprovação do projeto pelo Comitê de Ética em Pesquisa, CAAE 0315.0.045.000-10. ***Resultados*:** a análise dos dados revelou equilíbrio na realização dos diagnósticos no Piauí, apesar do Estado não contar com a identificação dessa doença pelo Teste do pezinho, encontrando-se ainda na Fase I, que abrange apenas triagem para Hipotireoidismo Congênito e Fenilcetonúria. ***Conclusão*:** ainda existem dificuldades no diagnóstico da doença, principalmente devido grande parte dos acometidos residirem no interior do estado. ***Descritores*:** Anemia Falciforme; Cuidado da Criança; Enfermagem.

RESUMEN

***Objetivo*:** caracterizar niños con anemia falciforme en los aspectos socioeconómicos y epidemiológicos. ***Método*:** estudio de naturaleza cuantitativa epidemiológica, realizado por medio de levantamiento en prontuarios de un Hospital Público en Teresina/PI, Nordeste del Brasil. Los sujetos fueron niños de 0 a 10 años internados por anemia falciforme, de diciembre de 2010 a marzo de 2011, cuando fueron realizadas las recolecciones de datos, después de la aprobación del proyecto por el Comité de Ética en Investigación, CAAE 0315.0.045.000-10. ***Resultados*:** el análisis de los datos reveló equilibrio en la realización de los diagnósticos en Piauí, a pesar del Estado no contar con la identificación de esa enfermedad por el Test del piecito, encontrándose aún en la Fase I, que abarca apenas selección para Hipotiroidismo Congénito y Fenilcetonuria. ***Conclusión*:** todavía existen dificultades en el diagnóstico de la enfermedad, principalmente debido en grande parte de los acometidos residir en el interior del estado. ***Descriptores*:** Anemia Falciforme; Cuidado del Niño; Enfermería.

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INTRODUCTION

In 1910, sickle cell disease was discovered by classical genetics through Dr. James Herrick, in Chicago. This was the first human molecular disease found in the blood of a black student, born in the West Indies, who had severe anemia, severe pain in joints and jaundice.[1,2] The term sickle cell disease encompasses a group of hereditary hemolytic anemia which have in common the presence of hemoglobin S within the red cell. It represents the most prevalent hereditary disease in the world, becoming more important in our country, in the regions that received massive contingent of Africans, because it was mainly introduced into the Americas by the slave trade from Africa, from XVI century.[3-5]

Sickle cell anemia is considered one of the hereditary hematologic diseases most prevalent in our environment but also has a significant epidemiological importance since it causes high morbidity and mortality if not diagnosed early and properly. It is considered a public health problem in Brazil, as our population has different ethnic and diverse degree of miscegeneration.[6,7] The origins of the S gene distribution in Brazil is quite heterogeneous, depending on Negroid or Caucasoid composition of the population. Thus, the prevalence of heterozygotes for Hb S is higher in the northern and northeastern regions (6 % to 10 %), while in the south and southeast prevalence is lower (2 % to 3 %).[8,9] The disease occurs in about 1 in every 500 African - American births and one in every 1000 to 1400 Hispanic-American births, with about 2 million Americans or 1 in 12 African Americans carry the sickle cell trait.[1.10]

The main feature of the disease is the ability of red blood cells, in certain circumstances losing their biconcave shape acquiring similar format with a distorted foice.[11] However, its clinical variability also worth mentioning: while some patients have a picture of great severity and are subject to numerous complications and frequent hospitalizations, others have a more benign course, in some cases, almost asymptomatic and contribution to this clinical variability both hereditary and acquired factors. Among the most important factors is acquired socioeconomic level with consequent variations in the qualities of food, infection prevention and medical care.[12] The hereditary/genetic factors are influenced by three characteristics: the haplotypes associated with the hemoglobin gene, the levels of fetal hemoglobin (Developmental inversely associated with severity) of the disease and concomitant alpha- thalassemia.[13]

Correct diagnosis is essential for proper treatment and counseling through laboratory confirmation of the result after two months old, together with clinical history and physical examination of the patient including the genotype of the biological parents.[5] In some Brazilian states, sickle cell anemia can be diagnosed shortly after birth through the "heel prick" but only from stage II, there is the aggregation of this disease, which is not a national reality whereas State of Piaui is still in phase I, not encompassing the hemoglobinopathy, which often complicates the implementation of an early diagnosis. Thus, considering that sickle cell disease although having treatment, it is still incurable and that improvements in living conditions directly influence the survival of the affected population, it is important to know the demographic profile of this population in order to achieve good results from the guidelines, particularly to family members, once the diagnosis is often performed later.

OBJETIVE

- To characterize children with sickle cell disease in terms of socioeconomic and epidemiological aspects.

METHOD

The epidemiological study was quantitative in nature. The study population consisted of hospitalized children who had sickle cell anemia as a cause of hospitalization in the period from December 2010 to March/2011, aged 0-10 years old, providing a sample of 21 patients.

The study was conducted in a public children's teaching hospital in Teresina-PI. A form containing demographic and epidemiological characteristics of the patient was used, which can be found in this handbook, therefore not necessitating contact and exposure of the patient.

Variables that relate to the distribution of children with sickle cell anemia according to demographic data, equivalent to age, sex, race/color, education and origin, presented in Table 1 were organized. Conferring epidemiology (Table 2), the following variables were studied: date of diagnosis, follow-up time for sickle cell disease, family history, housing conditions and sanitation.

The study was initiated after approval of the research project by Research Ethics Committee (CEP) of the Federal University of Piauí (UFPI) under CAAE 0315.0.045.000 – 10.

Children from 0 to 10 years old who had the disease and were hospitalized for complications related to sickle cell anemia were included in the study.

RESULTS AND DISCUSSION

The results of this study allowed us to identify the sociodemographic and epidemiological profile of children hospitalized with a diagnosis of sickle cell anemia in a Children Public Hospital of Teresina-PI. It is possible that certain facts may have underestimated the results, as the absence of necessary data in medical records, lack of parental knowledge to give relevant information to this study at the time of the interview, especially with regard to the presence of the disease among family or presence of family members carrying genes responsible for the onset of the disease, factors that have the power to limit some associations. The data led to two tables, containing, respectively: sociodemographic and epidemiological data.

In Table 1, the variables that concern the distribution of children with sickle cell anemia were organized according to sociodemographic data, equivalent to age, sex, race/color, education and origin.

Table 1. Distribution of children with sickle cell disease according to sociodemographic characteristics, from December 2010 / March 2011. Teresina (PI).

		n	%
Group age	6 – 10 months	03	14,30
	2 – 5 years old	11	52,40
	6 – 10 years old	07	33,30
Total		21	100,00
Sex	Male	12	57,00
	Female	09	43,00
Total		21	100,00
Race/color	White	01	5,00
	Black	02	10,00
	Asian	00	0,00
	Mulatto	15	71,00
	Indian	00	0,00
	Not applicable	03	14,00
Total		21	100,00
Education	Illiteracy	15	71,00
	Incomplete Elementar school	06	29,00
	Complete Elementary school	00	0,00
	Not applicable	00	0,00
Total		21	100,00
Origin	Urban Area of Teresina	07	33,30
	Countryside of Teresina	03	14,30
	Interior of Piauí State	10	47,60
	Other states	01	4,80
Total		21	100,00

The age range of these children were between two to five years old totaling 52.40%; six to ten years old totaling 33.30%, six to ten months totaling 14.30%. According to the age presented, education should include illiteracy (71%) and incomplete elementary education (29%), which was confirmed by the sample of children.

A study of 80 patients followed for at least one year at the Blood Center of Campinas, between the years 1986 and 1991 showed that the majority of patients with sickle cell anemia had the first complete or incomplete grade, followed by the second complete or incomplete grade and illiteracy. It is understood that the higher the level of education, more knowledge for the prevention and early treatment exist, knowledge that mainly involve parents.[4] If they have low education and restricted family income, possibly will lead to ignorance of the importance of care related to sickle cell anemia and may be an obstacle in the treatment of the child.[14] Literature data suggest that approximately 80% to 85% of patients with sickle cell disease have low education.15

In sex, 57% of the sample consisted of boys and 43% girls, which may be related to the small sample size. In relation to race they were 5% white, 10% black, 71% mulatto, still consider a percentage of 14% of children with unidentified color. It did not occur in the sample presence of Asian and indigenous race.

Studies in Brazil have shown a prevalence of Hb in newborns (NB) of approximately 4% to 5%, with no statistically significant difference between those with or without Hb NB regarding gender, weight and Apgar.[16] Because it is a genetic disease not related to sex, there are few publications addressing gender in sickle cell disease.[15] In race/ethnicity our study brought a predominance of mulatto color, which is explained by the great miscegenation of the Brazilian population, which received a large contingent of Africans at the time of its colonization, which causes the appearance of

the disease in a significant number of Brazilians

Taking into consideration the origin through a variable that consisted of specific objective of our research, we found as we expected, 47.6% of the sample comprised children in the state; 33.3% of the urban area; 14.3% of rural Teresina; and 4.8% from other states. It is noticeable that most affected are from the interior, often little knowledge of families that receive no guidance about the diseases that could affect the baby in the first months of life. The reality also shows a delay in Piauí, in relation to other states of Brazil, as the state is still in phase I of the Newborn Screening Program, not addressing screening for this disease.

Recognizing the prevalence of the disease, two important steps were taken by the Federal Government: the elaboration of the "Sickle Cell Anemia Program (FAP)" in 1996 and the creation of the "National Neonatal Screening Program (PNTN)" in 2001, Ordinance GM / MS No 822/01, which established the inclusion of tests for identification of sickle cell disease in routine examinations in all Brazilian newborns, known as "newborn screening test". Neonatal screening can save lives, depending on how parents understand this result for adherence to treatment plans and to avoid psychosocial complications, consisting of a preventive action that allows the diagnosis of various congenital or infectious, asymptomatic disease in the neonatal period. [17 8]

Among the congenital disease that the test identifies are hemoglobinopathies and among these, sickle cell disease, which is the most frequent and the most impactful because of its high prevalence and the severity of their clinical manifestations monogenic hereditary disease.[19] The screening test focuses in all three phases: phase I detects phenilcetonuria and hypothyroidism; Phase II phenilcetonuria, hypothyroidism and sickle cell disease; and Phase III phenilcetonuria, hypothyroidism, sickle cell disease and cystic fibrosis.

With regard to epidemiology (Table 2), the following variables were studied: date of diagnosis, follow-up, time for sickle cell disease, family history, housing conditions and sanitation.

Tabela 2. Distribuição de crianças com Anemia Falciforme, segundo dados epidemiológicos, no período de dezembro 2010/ março 2011. Teresina (PI), 2010/2011.

		n	%
Date of diagnosis	3 – 10 months	09	43,00
	2 – 5 years old	09	43,00
	Not applicable	03	14,00
Total		21	100,00
Follow up	2 – 4 years	15	71,50
	More than 8 years	02	9,50
	Not applicable	04	19,00
Total		21	100,00
Family history	Sickle cell Brothers	02	9,50
	Sickle cell Families	04	19,10
	Sickle cell mothermica	00	0,00
	Mother carrying gene	00	0,00
	Father carrying gene	00	0,00
	Family carrying gene	00	0,00
	Not applicable	15	71,40
Total		21	100,00
Housing conditions	Brick house	10	48,00
	Mud house	08	38,00
	Not applicable	03	14,00
Total		21	100,00
Sanitation	Presence of septic tank	10	48,00
	Absence of septic tank	08	38,00
	Not applicable	03	14,00
Total		21	100,00

The date of diagnosis revealed that in 43% cases, it was done a little later, when the child already was one-five year old, but an equivalent percentage of 43% showed a more accurate diagnosis, in which the discovery of disease was made up until ten months old, showing an advance in diagnosis. Only 3% of the sample had no date of discovery of the diagnosis.

It is known that early diagnosis favors taking preventive measures that can interfere positively in the treatment and outcome of the disease.[15] A study of children diagnosed through neonatal screening programs in Minas Gerais, between March/1998 to February 2005, showed that of 78 children that died, 42.3% of deaths occurred with up to two years old.[14] From this, it can be seen that the majority is at risk of death due to the disease. The objective diagnostics identify patients with sickle cell trait or disease requiring therapy or counseling.[20] Despite the fact that the gene for Hb S have been brought to Brazil with slavery, miscegenation diluted the

original African prevalence and now found the disease also apparently in Caucasians people.[21]

The advancement in the treatment of the disease and the consequent improvement in patient survival are intrinsically linked to how the family is welcomed and oriented from the diagnosis[18,22] Factors such as delayed diagnosis impact the prognosis of the disease that often it is only identified in times of crisis.

The clinical manifestations of the disease begin in the first year, extending throughout life and presenting large variability.[17] Clinical events depend on the age found: zero to five years old predominate: chronic anemia, painful crises, infections, dactylitis, splenic sequestration crisis, jaundice, stroke. In six to 12 years old predominate chronic anemia, painful crises, infections, brain, jaundice, ocular complications, gallstones stroke. From 13 years old predominate chronic anemia, painful crises, infections, stroke, eye complications, gallstones, leg ulcers, priapism, growth retardation, delayed secondary sexual characteristics, menarche and late first ejaculation.[11]

According to a study conducted with 80 subjects in the Hospital of UNICAMP, 69% had been diagnosed with sickle cell anemia within the last five years, 20% between five and 15 years and 11% for more than 15 years.[4] Thus, the date of diagnosis of children hospitalized to the children's Hospital of Piaui was similar, demonstrating to be late. However, compared to the same study, the diagnosis was much more providential, with dates of diagnosis identified, in most cases, within one to five years old, and 3 to 10 months old, which identifies a much more rapid diagnosis. As stated earlier, the sooner the diagnosis, the better the quality and length of life of these people.

The follow-up time in the hospital was 71.5% to an interval between two-four year of follow-up; 9.5% had just more than eight years in follow-up; and 19% had no record of this follow-up. Again, we see the consequences due to a delay in diagnosis that often is only held in times of disease complications, ending in the hospitalization of children.

In the family history, which aimed to evaluate family carriers of the disease or conducive to the development of disease genes, we found that: 9.5% had sickle brothers; 19.1% had sickle cell families and 71.4% did not have this knowledge about other family members. It is clear that the knowledge about the genome is still rare and often people are not even informed about the disease even having affected their children.

In children with sickle cell anemia, in general, parents are asymptomatic carriers of a single affected gene (heterozygous), producing HbA and HbS (AS), each transmitting the altered gene to the child, who thus receives the abnormal gene in double dose - homozygous SS. When a person receives an A gene and another S, becomes sickle cell trait (AS). The normal individual without the disease receives a pair of parent beta globin gene, which is responsible for the synthesis of hemoglobin making it AA.[6] If the child has the disease that is hereditary implies that if parents have not the disease, at least they have the gene cause. However, the study also shows an ignorance of parents about the disease process.

A survey of children and adolescents with sickle cell anemia treated at Hemope of Pernambuco/PE, in 2007, showed that out of ten families interviewed, seven had at least one sibling with sickle cell trait, then it is important to guide these families possibility of the birth of more children with the disease.[3]

Most of the housing conditions were the following: 48% live in brick house; 38% live in mud houses and 14% did not contain information regarding this aspect in the chart. In sanitation, there was an association with the item housing conditions, showing that 48% showed the presence of septic tank associated with brick house, while 38% had no septic tank, which is associated to residents in mud houses. Only 14% had no such information in the medical record.

The socioeconomic status of the population studied was low, considering that not many septic tank in their homes and a large proportion of children were living in mud houses. Studies showed a per capita income of 85% of the interviewed patients below one and a half minimum wage, in August 1992, a fact that remained the same in another study conducted in 2007, in which parents were in the age group between 22 and 40 years old and had only eight years of study, which compromised the family income, which ranged between one and two minimum wages.[3,4] Thus, the survey data in the medical records and based on comparisons with national literature found, we believe that the profile sickle cell anemia remains similar in different parts of the country.

CONCLUSION

Through this study, we have seen the importance of knowing the socio-demographic

and epidemiological profile of patients with sickle cell anemia and the need for a redefinition and improvement strategies developed by multidisciplinary teams for a more effective performance, as well as the disease can be diagnosed as early as possible for early treatment. It is also noticed that, despite this being a hereditary disease with the highest prevalence in the country, considered a major public health problem, studies are still scarce in the national literature. Therefore, it is expected the construction of other studies that seek to examine this subject.

It was exposed clearly in the study, children who were hospitalized for sickle cell anemia have similar profiles to each other, presenting unfavorable social conditions that will influence negatively the course of the disease. Therefore, solving the problem requires a number of changes in cultural policy mainly because as everyone knows their rights and the importance of identifying the disease early on, it will be easier to require the implementation of this diagnosis in their localities, reducing complications of this disease in the country.

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