RELATO DE CASO

SICKLE CELL ANEMIA IN QUILOMBOLAS: A LITERATURE REVIEW

ANEMIA FALCIFORME EM QUILOMBOLAS: UMA REVISÃO DE LITERATURA

Renan Pires Ferreira Borges[1], Rafael Felipe Carvalho Canutto[1], Kassita Alvarenga Gomes[1], Gustavo Carneiro Resstel[1], Guilherme Pansani Silva[1], Carlos Alberto Rodrigues Junior[1], Fellipe Camargo Ferreira Dias[2], Virgílio Ribeiro Guedes[3].

ABSTRACT

Introduction: Sickle cell disease is a generic term that encompasses a set of hematological disorders linked to structural changes in blood cells, leading to the production of an abnormal hemoglobin called HbS, the 'S' being derived from English sickle. The objective of this study was to discuss the profile of sickle cell anemia in quilombola communities by reviewing the existing literature in order to provide updated data to professionals dealing with this population. Development: The prevalence of HbS heterozygotes in Brazil is higher in the North and Northeast regions, between 6% and 10%, and considerably lower in the South and Southeast regions, with prevalence between 2% and 3%. In this study, it was found that 6.6% to 11.5% of the population of quilombolas in the state of Tocantins had abnormal hemoglobins. Conclusion: Few studies evaluate the prevalence of hemoglobinopathies in quilombola communities. Since quilombolas usually live in isolated communities of society, the chances of marriages between individuals with sickle cell trait are greater than in the general population, thus increasing the probability of being born to individuals affected by the disease.

Keywords: Sickle cell disease, Quilombolas, Epidemiology.

RESUMO

Introdução: Doença falciforme é um termo genérico que engloba um conjunto de desordens hematológicas ligadas a alterações estruturais das células sanguíneas, levando à produção de uma hemoglobina anormal denominada HbS, sendo o 'S' derivado do inglês sickle. O objetivo deste estudo foi abordar o perfil da anemia falciforme nas comunidades quilombolas por meio de revisão da literatura existente, a fim de fornecer dados atualizados aos profissionais que lidam com essa população. Desenvolvimento: No Brasil, a prevalência de heterozigotos HbS é maior nas regiões do Norte e Nordeste, entre 6% e 10%, e consideravelmente menor nas regiões Sul e Sudeste, com uma prevalência entre 2% e 3%. Neste estudo, encontrou-se que 6,6% a 11,5% da população de quilombolas no estado do Tocantins apresentaram hemoglobinas anormais. Conclusão: Poucos estudos avaliam a prevalência de hemoglobinopatias em comunidades quilombolas. Visto que os quilombolas costumam viver em comunidades isoladas da sociedade, as chances de ocorrerem casamentos entre indivíduos com traço falciforme, tornam-se maiores que na população em geral, aumentando assim a probabilidade de nascerem indivíduos acometidos pela doença.

Palavras-chave: Doença falciforme, Quilombolas, Epidemiologia.

ACESSO LIVRE

Citação: Borges RPF, Canutto RFC, Gomes KA, Resstel GC, Silva GP, Rodrigues Junior CA, Dias FCF Guedes VR (2018) Sickle cell anemia in quilombolas: a literature review. Revista de Patologia do Tocantins, 5(1): 34-37.

Instituição: [1]Acadêmico(a) de Medicina, Universidade Federal do Tocantins, Tocantins, Brasil; [2]Médico, Mestrando em Ensino em Ciências e Saúde, Universidade Federal do Tocantins, Tocantins, Brasil; [3]Docente, Médico de Medicina da Família e Comunidade, Universidade Federal do Tocantins, Tocantins, Brasil.

Autor correspondente: Renan Pires Ferreira Borges; renanpfb@gmail.com

Editor: Guedes V. R. Medicina, Universidade Federal do Tocantins, Brasil.

Publicado: 16 de abril de 2018.

Conflito de interesses: os autores declararam que não existem conflitos de interesses.

INTRODUCTION

Sickle cell disease (SCD) is a generic term that encompasses a set of hematological disorders linked to structural changes in blood cells, leading to the production of an abnormal hemoglobin called HbS, the 'S' being derived from English sickle[1,2]. Sickle cell disease, as well as sickle cell disease, is determined by replacing the glutamic amino acid with valine in the sixth codon of the betaglobin chain[3,4].

SCD is the most common hereditary hematologic disorder in Brazil and throughout the world[5]. In 2010, it was estimated that 200,000 infants are born annually with the disease in Brazil[6]. Although SCD is not a biological marker for race, it can be considered a marker for ancestry from a geographic location where malaria is or was prevalent. Given that the biology of sickle cell stems from the endemicity of malaria and the subsequent protective and genetic response, and as a result is not attached to any one ancestral or racial group, it could be a disease that represents the Brazilian lore of a geographically, culturally, and racially diverse and integrative nation[7-10]. Instead, the Brazilian state has assigned SCD almost exclusively to Afro-Brazilians[11].

According to Bender & Hobbs[12], SCD is characterized by vaso-occlusive events and chronic hemolytic anemia. Occlusion episodes produce tissue ischemia, leading to acute and chronic pain, as well as damage to various organs such as bones, lungs, liver, kidneys, brain, eyes, and joints. Pain and / or swelling in the hands or feet in children or newborns is often the first symptomatology of SCD[12].

The disease mainly affects the black population, which mostly lives in situations of poverty and social vulnerability[13]. Researcher Kikuchi estimates that, in 2003, about 85% of Brazilian adult sickle cell patients had low schooling. The few patients with sickle cell anemia who can enter the labor market are submitted to manual services, considerably aggravating their physical situation[12,14]. According to Cavalcante[15] it is perceived that students with SCD are not fully served in their needs and rights. Often, they are injured in their school day because of their absences due to the symptoms of the disease. What has been found is that often these students do not have the opportunity to enjoy satisfactory learning because they are not well matched to their cognitive, personal and school needs[15].

Since quilombolas live in communities that are generally isolated from society, it is perceived that the chances of marriages occur between individuals with sickle cell trait become large, thus increasing the probability of being born sick individuals. Thus, the information, genetic orientation and genetic counseling of quilombolas are of great relevance, since it offers the opportunity of family planning and reproductive decision in the presence of genetic risk[16,17].

The objective of this study was to discuss the profile of sickle cell anemia in quilombola communities by reviewing the existing literature in order to provide updated data to professionals dealing with this population.

DEVELOPMENT

SCD is an important political site in which race, citizenship, biological determinism, ancestry, and health are contested. Historically associated with Blackness[18,19], SCD was chosen by activists as the quintessential representation of a disease that set Black Brazilians apart both biologically and culturally[11]. It is considered an ethno-racial disease due to its geographical origin and genetic etiology, and by prevalence statistics in the black population. However, some care should be taken in the classification of the disease as such, which implies a process called by Laguardia[20] as a racialization of SCD[4].

Hb S has been studied not only in patients with sickle cell disease, but also in patients with this variant in heterozygosity (Hb AS). In the case of quilombola communities, because they are localities formed mainly by Afrodescendants, they present greater geographic isolation and, consequently, the existence of consanguineous marriages, therefore, the importance of this investigation. The lack of knowledge about sickle cell disease and its clinical disorders often reflects in the late diagnosis of hemoglobin, which is an important facilitator for the dissemination of Hb S[21].

A study conducted by Dias et al[22] aimed to characterize the network of social and affective support and to know the coping strategies used by relatives of people with sickle cell disease. The results showed an average of 28 contacts in the support network of this sample, most of whom were considered satisfactory contacts. Focusing on the problem was the most used by family members. Thus, support network knowledge and coping strategies of sickle cell family members allows the planning of more effective psychosocial interventions promoting quality of life for this population[22].

Another study, conducted to determine the prevalence of HbS among Asian, Afro and Euro-Brazilian individuals from a blood bank in Curitiba, showed that the overall prevalence of HbS in the study population was 0.9%, being 0 % among Asian-Brazilians, 2.7% among Afro-Brazilians and 0.7% among Euro-Brazilians. There was a positive association, statistically significant, for the sickle cell trait in people of African descent. Thus, it demonstrated higher rates of sickle cell trait in Afro-Brazilians, which corroborates data published in other Brazilian regions and states[23].

According to Cançado and Jesus[5], the heterogeneity of the distribution of SCD in Brazil may be related to the constitution of the local population, and the disease is more incident in regions constituted by large contingents of Afro-Brazilians. Thus, the prevalence of HbS heterozygotes is higher in the North and Northeast regions, between 6% and 10%, and considerably lower in the South and Southeast regions, with prevalence between 2% and 3%[4].

A study conducted by Soares[24] had the objective of investigating the presence of variant hemoglobins in 15 quilombola communities in the state of Piauí. A total of 1,239 samples were analyzed, in which the hemoglobins were sorted by high performance liquid chromatography (HPLC). A questionnaire was applied regarding gender, ethnicity and consanguinity of the populations. Of the 1,239 samples, 5.4%

showed AS, and sickle diseases SS and SC were found in 0.8% of total hemoglobins AC, AD and DD. Of the 1,069 black people, 84 had hemoglobin changes; of these, 34 were males and 53 females. There were 13 consanguineous marriages among the 84 hemoglobin alterations[24].

Work carried out by Souza[25] made the screening of hemoglobin S and C and the study of the social profile of four quilombola communities. The study was developed in the quilombola communities of Malhadinha, Córrego Fundo, Curralinho do Pontal and Manoel João, municipality of Brejinho de Nazaré / Tocantins. Eleven samples (6.6%) with variant hemoglobins were identified: eight (4.8%) with sickle cell trait and three with hemoglobin C (1.8%). Social data were obtained through an interview with representatives of 48 families, collecting information on family income, age, sex and occupational activity of each individual. The study showed that these communities are formed predominantly by adults and the elderly, with monthly income greater than a minimum wage, for 50% of the families interviewed. Although the percentage of hemoglobins S and C found in the communities is within that observed for several regions of Brazil, the lack of information on the disease and social aspects can increase the number of individuals with SCD in the municipality or neighboring areas[25].

Another study aimed to verify the incidence of hemoglobins of African descent (HbS and HbC) in quilombola communities in the state of Tocantins, Brazil. The blood of quilombolas was collected in 14 communities of the State; the screening was performed in cellulose acetate electrophoresis (pH 8.6), and those with altered standard were submitted to high performance liquid chromatography, and the gender and age of the individuals sampled were recorded. The analysis of the results showed that, of the 822 quilombolas investigated, 95 presented abnormal hemoglobins, being 0.5% with sickle cell disease (HbSS); 5.7% trait for hemoglobin S (HbAS); 4.9% trait for hemoglobin C (HbAC); 0.2% with increased fetal hemoglobin; 0.1% with increased hemoglobin A2; and 88.4% with normal hemoglobin (HbAA). HbSS was observed in the infant and adolescent age group and HbAS and HbAC in all age groups. Regarding sex, it was not possible to suggest the maternal effect for HbS due to the larger number of males with this genetic information. In this study, the incidence of HbS and HbC, observed in quilombola communities, was within the expected range for the Northern Region of Brazil. However, the high prevalence of SCD and the high frequency of sickle cell trait in some of the studied communities stand out, with special attention to the southern region of the State[26].

CONCLUSÃO

Since quilombolas usually live in isolated communities of society, the chances of marriages between individuals with sickle cell trait are greater than in the general population, thus increasing the probability of being born to individuals affected by the disease.

Few studies evaluate the prevalence of hemoglobinopathies in quilombola communities. In this study, it was found that 6.6% to 11.5% of the population of quilombolas in the state of Tocantins had abnormal hemoglobins.

REFERENCES

1. Felix AA, Souza HM, Ribeiro SBF. Aspectos epidemiológicos e sociais da doença falciforme. Revista Brasileira de Hematologia e Hemoterapia. 2010; 32: 203-208.
2. Neto GCG, Pitombeira MS. Aspectos moleculares da anemia falciforme. Jornal Brasileiro de Patologia e Medicina Laboratorial. Rio de Janeiro. 2003; 39(1): 51-56.
3. Assis ES. Estudo das síndromes falcêmicas em comunidade quilombola, Sergipe/Brasil. Programa de Pós-graduação em Saúde e Ambiente. Universidade Tiradentes. Aracaju. 2010.
4. Lopes WSL. Impacto social da doença falciforme em comunidades quilombolas de Paracatu, Minas Gerais, Brasil. Faculdade De Ciências E Tecnologia. Tese de mestrado. 2013: 83.
5. Cançado RD, Jesus JA. A doença falciforme No Brasil. Rev. Bras. Hematol. Hemoter. 2007; 29 (3).
6. Jesus JA. Doença falciforme no Brasil. Gaz. Médica Bahia. 2010; 3.
7. Bala P. Biomedicine as a Contested Site: Some Revelations in Imperial Contexts. Lexington Books, Lanham, MD. 2009.
8. Lima NT. Public health and social ideas in modern Brazil. Am. J. Public Health. 2007; 97 (7): 1168-1177.
9. Peard JG. Tropical disorders and the forging of a Brazilian medical identity, 1860-1890. Hispanic Am. Hist. Rev. 1997; 77 (1): 1.
10. Stepan N. Beginnings of Brazilian Science: Oswaldo Cruz, Medical Research and Policy. Science History Publications, New York. 1976: 1890-1920
11. Creary MS. Biocultural citizenship and embodying exceptionalism: Biopolitics for sickle cell disease in Brazil, Social Science & Medicine. 2017; 1-9.
12. Bender MA, Hobbs W. Sickle cell disease. University of Washington. Seattle. 2003: 1993-2013.
13. Kikuchi BA. Diáspora Africana e anemia falciforme. In: Racismos contemporâneos (Org.) Ashoka Empreendedores sociais e Takano Cidadania. Rio de Janeiro. 2003.
14. Guimarães TMR, Miranda WL, Tavares MMF. O cotidiano das famílias de crianças e adolescentes portadores de anemia falciforme. Revista Brasileira de Hematologia e Hematoterapia. 2009; 21(1): 9-14
15. Rodrigues WCC, Seibert CS, Ferreira da Silva KL. Um olhar sobre a formação do aluno com doença falciforme. In: Revista Desafios 4. 2017; 1: 86-94
16. Mingroni-Netto RC, Auricchio MTBM, Vicente JP. Importância da Pesquisa do Traço e da Anemia Falciforme nos Remanescentes de Quilombos do Vale do Ribeira-SP. In: Saúde nos Quilombos/ Editado por Anna Volochko e Luís Eduardo Batista – Instituto de Saúde – SESSP, São Paulo: GTAE – SESSP, 2009; 169-177.
17. Meneses RCT. Intervenção Pedagógica Em Saúde, Aconselhamento Genético E Autocuidado Com A Saúde E Alimentação Para Comunidades Quilombolas. Universidade Tiradentes. Programa De Pós-Graduação Em Saúde E Ambiente. Tese de mestrado. 2013; 1-83.
18. Tapper M. In the Blood: Sickle Cell Anemia and the Politics of Race. University of Pennsylvania Press. 1999.
19. Wailoo K. Dying in the City of the Blues: Sickle Cell Anemia and the Politics of Race and Health. UNC Press Books.2001.

20. Laguardia J. No fio da navalha: anemia falciforme, raça e as implicações no cuidado à saúde. Estudos Feministas. Escola Nacional de Saúde Pública (FIOCRUZ). Florianópolis. 2016; 14(1): 243-262.
21. Andrade SP, et al. A distribuição da hemoglobina S em três comunidades quilombolas do estado do Tocantins-Brasil. Scientia Amazonia. 2015; 4(1): 10-20.
22. Dias TL, Leite LLG. Rede de apoio social e afetivo e estratégias de enfrentamento na doença falciforme: um olhar sobre a pessoa e a família. Psicologia em Revista, Belo Horizonte. 2014; 20(2): 353-373.
23. Lidani KCF, Barros RF, Bovo F. Relationship between the prevalence of hemoglobin S and the ethnic background of blood donors in Paraná state. J Bras Patol Med Lab. 2015; 51(4): 212-217.
24. Soares LF, et al. Prevalência de hemoglobinas variantes em comunidades quilombolas no estado do Piauí, Brasil.Ciênc. saúde coletiva [online]. 2017; 22 (11): 3773-3780.
25. Souza LO, et al. Triagem das hemoglobinas S e C e a influência das condições sociais na sua distribuição: um estudo em quatro comunidades quilombolas do Estado do Tocantins. Saude soc.[online]. 2013; 22(4): 1236-1246.
26. Figueredo TA, et al. Hemoglobinas de origem africana em comunidades quilombolas do estado do Tocantins, Brasil. Rev Pan-Amaz Saude [Internet]. 2017; 8(1): 39-46.