Hemophiliacs profile provided of a patient association

Perfil dos hemofílicos provenientes de uma associação de pacientes

Perfil de hemofílicos de una asociación de pacientes

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RESUMO

Objetivo: traçar o perfil epidemiológico de hemofílicos vinculados a uma associação de pacientes do estado de Goiás, Brasil. **Método:** pesquisa transversal realizada com indivíduos do gênero masculino acima dos 18 anos de idade. Catorze participantes foram submetidos a uma ficha de avaliação contendo questões sociodemográficas e clínicas. O presente estudo utilizou análises de frequências para descrição da amostra. Para as variáveis quantitativas rodou-se o teste de normalidade de Shapiro Wilk. **Resultados:** a média de idade encontrada foi de 32,64 anos ± 9,32. A maior parte dos indivíduos residia a menos de 30 km do centro de tratamento e apresentaram a forma grave da doença. Em relação às comorbidades, um indivíduo apresentou inibidor do fator de coagulação e as infecções virais estiveram ausentes em 71,4% dos participantes, em um período que não havia controle antiviral. As hemorragias articulares predominantes foram observadas no cotovelo e joelho e o hematoma muscular esteve presente em 50% da amostra. O tratamento mais utilizado pelos participantes foi a profilaxia secundária. **Conclusão**: a partir da caracterização dos pacientes hemofílicos cadastrados em uma associação é possível compreender mais sobre a patologia em estudo, demonstrando que as infecções virais se constituem em importantes comorbidades adquiridas por hemofílicos adultos.
Descritores: Hemofilia A; Hemofilia B; Adulto; Perfil de Saúde.

ABSTRACT

Objective: to trace the epidemiological profile of hemophiliacs linked to a patient association in the state of Goiás, Brazil. **Method:** cross-sectional research conducted with male individuals over 18 years old. Fourteen participants were submitted to an evaluation form containing sociodemographic and clinical questions. The present study used frequency analysis to describe the sample. For quantitative variables, the Shapiro Wilk normality test was run. **Results:** the average age found was 32.64 years ± 9.32. Most individuals lived less than 30 km from the treatment center and had a severe form of the disease. Regarding comorbidities, one individual had a coagulation factor inhibitor and viral infections were absent in 71.4% of the participants, in a period when there was no antiviral control. The predominant joint hemorrhages were observed in the elbow and knee and muscle hematoma was present in 50% of the sample. The most used treatment by the participants was secondary prophylaxis. **Conclusion:** from the characterization of hemophiliac patients registered in an association, it is possible to understand more about the pathology under study, demonstrating that viral infections are important comorbidities acquired by adult hemophiliacs.
Descriptors: Hemophilia A; Hemophilia B; Adult; Health Profile.

RESUMEN

Objetivo: rastrear el perfil epidemiológico de hemofílicos vinculados a una asociación de pacientes en el estado de Goiás, Brasil. **Método:** investigación transversal realizada con varones mayores de 18 años. Se enviaron catorce participantes a un formulario de evaluación conteniendo preguntas sociodemográficas y clínicas. El estudio utilizó análisis de frecuencia describiendo la muestra. Para las variables cuantitativas se ejecutó la prueba de normalidad de Shapiro Wilk. **Resultados**: la edad promedio fue de 32,64 años ± 9,32. La mayoría de las personas vivían a menos de 30 km del centro de tratamiento y tenían una forma grave de la enfermedad. En cuanto a las comorbilidades, un individuo tenía un inhibidor del factor de coagulación y las infecciones virales estaban ausentes en el 71,4% de los participantes, en un período en el que no hubo control antiviral. Las hemorragias articulares predominantes se observaron en el codo y la rodilla y el hematoma muscular estuvo presente en el 50% de la muestra. El tratamiento más utilizado por los participantes fue la profilaxis secundaria. **Conclusión:** a partir de la caracterización de los pacientes hemofílicos, es posible conocer más sobre la patología en estudio, demostrando que las infecciones virales son importantes comorbilidades adquiridas por hemofílicos adultos.
Descriptores: Hemofilia A; Hemofilia B; Adulto; Perfil de salud.

ORIGINAL

Introduction

Hemophilia is characterized as a male-linked hereditary coagulopathy. Its transmission is made by the X chromosome, manifesting itself in most cases in males. The term hemophilia is used to indicate the levels of deficiency of coagulation factor VIII (hemophilia A) or factor IX (hemophilia B), being observed in the first one frequency and 85% of cases and in the second with 15%.[1]

The incidence of hemophilia A is approximately 1:10,000 to 1:30,000 births, while hemophilia B is 1:30,000 to 1:50,000. Both course with the same clinical presentation, making it indispensable to diagnose the activity of specific coagulation factors, factor VIII and factor IX, for differentiation between them.[2]

Comorbidity can be classified as mild, moderate or severe as mild, moderate or severe according to symptoms and the degree of deficiency of factors VIII or IX. In severe or severe deficiency, hemorrhagic intake is higher and has a concentration of factors below 1%. It is manifested by spontaneous bleeding in the joints or muscles. When the deficiency is moderate, the concentration of the factor is 1% to 5%, generating occasional spontaneous bleeding, prolonged bleeding with less trauma.[3] When factor levels are between 5% and less than 40%, it is characterized by mild hemophilia. There is usually no presence of hemarthrosis and other spontaneous bleeding, although problematic bleeding may arise in cases of surgery or injury.[3]

Hemorrhages occur mainly on the form of hematomas and hemarthrosis, the second being repeated and when left untreated is associated with joint destruction, called hemophilic arthropathy, resulting in chronic pain, joint deformities and severe functional impotence.[4] Hemarthrosis consists of the extraleakage of blood into the joint or into the synovial cavity.[5] Other forms of bleeding may occur on the form of hematuria, epistaxis, melena/hemathemesis intracranial hemorrhage and retroperitol bleeding.[4]

Despite changes in the locomotor system in patients with hemophilia, the disease is usually not life-threatening. However, the sequelae resulting from this process may generate disabilities.[6]

The treatment of hemophilia is carried out under different modalities, demand and prophylactic. Demand treatment refers to that which occurs after an episode of bleeding. The intensity and duration of treatment will depend on the location and severity of bleeding. This treatment was the only one available in Brazil until December 2011. Prophylactic treatment refers to that which occurs prior to the development of the bleeding episode, and may be primary, when initiated before the second hemarthrosis or before the first 2 years of age, or secondary, when it does not meet the criteria for primary prophylaxis and the patient already shows signs of joint sequelae. Prophylactic treatment is indicated in severe haemophilia and has as main objective to prevent repeat hemarthrosis, which can cause target joints and permanent functional deformities.[4]

The transmission of infectious agents, especially hepatitis agents, was due to the transfusion of blood components and blood products not submitted to an adequate process of viral inactivation. The emergence of acquired immunodeficiency syndrome (AIDS) in 1981, caused by HIV, directly affected the hemophilic population due to its treatment.[7]

One of the most fearsome complications of hemophilia patients refers to the development of inhibitors, which are antibodies directed against factors VIII or

IX infused. In this case, affected patients do not respond to deficient factor infusion and present hemorrhagic episodes of difficult control.[4]

Knowing that hemophilia has no ethnic or geographical limits and that Brazil is classified as the third country with the highest hemophiliac rate, and India is first, followed by the United States.[8]

In this context, this study aims to describe sociodemographically and clinically the profile of adult hemophiliac patients from an association of patients from the state of Goiás, Brazil.

Method

This is a cross-sectional research. Data collection was performed at the Hemophiliacs Association of the State of Goiás (AHEG). The sample was for convenience and data were collected in November and December 2015.

Hemophiliac individuals linked to AHEG, male, literate, over 18 years old, were invited to participate in the research. The Informed Consent Form (TCLE) was given, which all participants formally signed. Hemophiliacs with cognitive and indigenous impairment were excluded.

For the sociodemographic collection, an evaluation form was used prepared by the research team. In the form there were questions for sociodemographic and clinical characterization of patients. The sociodemographic variables were: age, race/color, marital status, education, private health plan, employment status, receipt of government benefit/financial aid, practice of physical activity and distance between residence/place of treatment. The clinical variables were: types of hemophilias, clinical severity, inhibitors, viral infections, types of treatments, use of joint prosthesis, site of more frequent hemorrhages.

The present study used frequency analyses to describe the sample. For the quantitative variables, the Shapiro Wilk normality test was rotated. All statistical analyses were performed by the Statistical Package SPSS 20.0.

The project was approved by the Ethics Committee on Research with Human Beings of the Federal University of Mato Grosso do Sul (UFMS), through plataforma Brasil, in accordance with Resolution No. 466 of 2012, of the National Health Council, and being approved under opinion no. 1,300,316.

Results

Sociodemographic and clinical characterization of the study sample was performed, according to tables 1, 2 and 3.

Table 1. Sociodemographic information of hemophiliacs evaluated in this study. Federal District, 2015.

Variable	%
Age range (years)	**% (n=14)**
20-29	50,0
30-39	28,6
40-49	7,1
50-59	14,3
Mean age	32,64±9,32

Marital Status	**% (n=14)**
Married	50
Single	50
Color/Race	**% (n=14)**
Branco	28,6
Pardo	64,3
Negro	7,1
Education level	**% (n=14)**
1st Degree Incomplete	7,1
1st Degree Complete	14,3
2nd Degree Incomplete	14,3
2nd Grade Complete	35,7
3rd Degree Incomplete	14,3
3rd Degree Complete	14,3
Health Plan	**% (n=14)**
Yes	42,9
No	57,1
Government Benefit/ Financial Aid	**% (n=14)**
Yes	42,9
No	57,1
Distance from residence to place of treatment	**% (n=14)**
Less than 30Km	71,4
More than 30Km	28,6

Table 2. Profile of Work Activity and Physical Activity of Hemophiliacs. Federal District, 2015.

Work Activity	**% (n=14)**
Yes	42,9
No	57,1
Physical Activity	**% (n=14)**
Yes	42,9
No	57,1

Table 3. Characterization of hemophilia, its probable symptoms and treatments.

Type of Hemophilia	**% (n=14)**
A	92,9
B	7,1
Gravity	**% (n=14)**
Take	14,3
Moderate	21,4
Serious	64,3
Inhibitor	**% (n=14)**
Yes	7,1
No	92,9

Virus Infection	% (n=14)
No	71,4
Hepatitis C	14,3
Hepatitis B and C	7,1
Hepatitis C and HIV	7,1
Joint Prosthesis	**% (n=14)**
Yes	14,3
No	85,7
Type of Treatment	**% (n=14)**
P1	21,4
P2	57,1
DEM	14,3
FEIBA*	7,1

*Tratamento para inibidores dos fatores de coagulação.

Discussion

The mean age of the individuals was 32.64 ± 9.32. The age group from 20 to 29 years had a higher prevalence, representing 50% of the sample. Similar results were found by Silva9 with 175 adult hemophiliacs (18 to 64 years), with a mean age of 31.44 years. According to the Profile of Hereditary Coagulopathies8, the highest prevalence of hemophilias occurs in the age group 20 to 29 years, corroborating the results found in the present study.

Regarding marital status, the sample was divided into 50% married individuals and 50% single individuals. In the study by Garbin et al., 60% of the individuals were single.10 Such data can be explained by the difficulties in facing the disease and its consequences, such as the physical limitations resulting from bleeding and the impossibility of working. In addition, patients describe the difficulty in being accepted when they claim to be hemophiliacs and, especially, when they reveal that they have some infectious disease. On the other hand, Caio et al., in their sample, observed that 83% of the individuals were married or ameded and that the constitution of a family represents an important point of support for hemophiliacs.[11]

In this sample, 64.3% of the individuals were brown, 28.6% white and 7.1% black. Garbin et al. found divergent results, in which most individuals were white and a small portion were mulattos and blacks.[10] However, it should be noted that according to Manno, there is no ethnic distinction for the disease.[12]

Regarding the level of education, there was a predominance of individuals with complete high school (35.7%). According to Santos and Ferraz, 35.3% of hemophiliacs had completed high school, and it is important to highlight that this sample has an age group of 23.6 years.[13] In the study by Caio et al., the age distribution and access to a better level of education do not differ between patients with hemophilia and their siblings who do not have the disease.[11]

In 2013, 15,000 patients with the disease were assisted by the public health network (they received medicines through the SUS, including those who had health insurance and insurance or who went to the private health system). Of this total of patients, 10,464,000 were registered as hemophiliacs A and B.[4] Nevertheless, the results of the present study show that 57.1% of hemophiliacs

did not have health insurance. Still, 57.1% did not receive any kind of benefit. However, unlike the data from this research, Almeida et al. reported that 72.7% of patients received some kind of government benefit.[14]

Regarding the distance from the Treatment Center and the residence, it was found that most individuals (71.4%) resided less than 30 km from their place of treatment, which would facilitate emergency care in case of bleeding. It is easy to conclude that the individual with hemophilia may require, at any time, an urgent replacement of coagulation factor, high operational cost therapy and generally available in few specialized centers. This fact creates a peculiar psychosocial situation for the patient with hemophilia, with the constant threat of unexpected bleeding and dependence, in terms of geographical distance and availability, of a specialized treatment center.[11]

In the present study, we found an index of 42.9% of individuals who performed physical activity and 42.9% performed work activity. Divergent results were presented by Nunes et al. in their research with 23 patients, of whom 69% did not perform physical activity and 82.6% did not work.[15]

For Santos and Ferraz, in an epidemiological survey with 17 hemophiliac individuals, only one performed walks as physical exercise and eight did not work.[13] Andery et al. showed that the practice of physical activity helps in the treatment of hemophilia, since physical exercises improve the balance of the musculature, stabilization of the joint, preventing possible bleeding and offering a better social interaction.[16] However, it is necessary to avoid intense contact activities.

According to the study by Nunes et al., the most frequent type of hemophiliacs type A, with 91.3% and 8.7% hemophiliacs of type B.15 Tavares et al., found 97.1% of cases of hemophilia A, in a population of 102 participants.[17] The present study corroborates the literature, which highlights hemophilia A as more prevalent than hemophilia B, with 92.9% and 7.1% respectively.[1]

Regarding severity, it was observed in this study that most cases were severe (64.3 %), followed by 21.4% moderate and 14.3% mild cases. Divergent results were found by Santos et al., where they verified the predominance of the moderate type with 22%, severe (21%) and mild (16%). However, there were 41% (63 medical records) with indetermination of clinical characterization.[18]

In the present study, of the 14 patients questioned, 92.9% (n=13) did not present inhibitors. Affected patients with the development of 12 inhibitors are generally those affected by severe hemophilia A, manifesting poor response to the usual treatment or increased frequency and/or severity of bleeding.2 The incidence of inhibitor development is about 5-15% of patients with hemophilia A and about 3% in patients with hemophilia B, with the results of this study below these rates.[19]

Among the patients, 28.6% had some type of viral infection by the use of contaminated blood products, with 14.3% acquiring Hepatitis C, 7.1% hepatitis B and C and the others, 7.1% hepatitis C and HIV. In the 1980s hemophiliac patients were much more susceptible to Hepatitis B, Hepatitis C and HIV viruses, as criteria for evaluating contaminated blood components were not met.[20]

In this study, we found a higher rate of bleeding in the elbow (64.3%), followed by knee (57.1%), muscle hematoma (50%), ankle (42.9%) and nasal (35.7%). In line with the study by Wisniewshi and Kluthcovsky[21], conducted in 2008, shows that the most frequent complication was elbow hemarthrosis with 27.6%. According to Rodriguez Merchan, joints such as knees, elbows and ankles

account for 80% of hermartroses in patients with severe hemophilia A.[22] Since hemophilia is a chronic disease characterized by the involvement of the musculoskeletal system, it generates the restriction of joint mobility and the development of muscle deformities, thus limiting the functionality of these individuals.[22]

Of the 14 individuals, 14.3% (n=2) used joint prosthesis, one with severe hemophilia A and the other moderate hemophiliac B. In the study by Almeida et al., however, they found that of 33 individuals, three had prostheses and all presented the severe form of the disease.[14]

In this study, it was observed that 57.1% use the treatment of secondary prophylaxis, in which according to Srivastava et al. consists of regular and continuous replacement (minimum of 45 weeks per year) of the coagulation factor, where treatment begins after two or more joint bleeding.[3] Corroborating the Profile of Hereditary Coagulopathies, in the state of Góias, between the years 2013 to 2015, 150 individuals used secondary prophylaxis, that is, the treatment most used in the present study.[8]

Conclusion

The research showed that the sample of adult patients was born in a period of great contagion due to infectious diseases, through blood products, however, the vast majority (71.4%) was not contaminated, which would increase the morbidity and mortality of patients. Another point to be highlighted is that the majority of participants (71.4%) lies close to its place of treatment, facilitating access and care in emergency cases.

The present study had limitations due to the small number of hemophiliac participants, however it was possible to draw a profile of adult patients linked to the HemophiliacS Association of the State of Goiás (AHEG), a profile similar to that present in the current literature in aspects such as the regions of the body where hemorrhages are more common, demonstrated in the study by Wisniewshi and Kluthcovsky and in the research of Rodriguez Merchan , but divergent in other topics, such as the proportion, presented by Nunes et al, of patients with the disease who practice physical activity and perform work activity.

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