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RESEARCH

Signs and symptoms in Gaucher Disease: priority nursing diagnoses

Sinais e sintomas na Doença de Gaucher: diagnósticos de enfermagem prioritários

Señales y síntomas de la Enfermedad de Gaucher: diagnósticos prioritarios de enfermería

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ABSTRACT

Objective: Identify the signs and symptoms of patients with Gaucher Disease, inferring possible priority nursing diagnoses. **Method**: Cross-sectional study, developed in a specialized laboratory, between 2013 and 2015. The sample (n = 91) comprised the records of patients with genetic diagnosis for Gaucher Disease. The study respected research norms. **Results**: Prevalence of female sex (57.1%), age at diagnosis between 0 and 10 years, and origin from the Southeast Region of Brazil were prevalent. Hematologic changes, bone pain, hepatomegaly, splenomegaly, and fatigue were the most recurrent signs and symptoms. The inferred diagnoses for the studied population were: Risk for bleeding; Fatigue; Chronic pain and Acute pain; Impaired physical mobility; Imbalanced nutrition: less than body requirements; and Risk for Developmental Delay. **Conclusion**: The establishment of Priority Nursing Diagnoses based on signs and symptoms makes it possible to achieve expected outcomes for each individual in the care context.

Descriptors: Gaucher Disease; Cerebrosid Liposidosis Syndrome; Signs and Symptoms; Nursing Process; Nursing Diagnosis.

RESUMO

Objetivo: Identificar os sinais e sintomas de pacientes com Doença de Gaucher, inferindo os possíveis diagnósticos de enfermagem prioritários. **Método**: Estudo transversal, desenvolvido em laboratório especializado, entre 2013 e 2015. A amostra (n=91) foi constituída dos registros de pacientes com diagnóstico genético para Doença de Gaucher. O estudo respeitou normas de pesquisa. **Resultados**: Foram prevalentes o sexo feminino (57,1%), faixa etária ao diagnóstico entre 0 e 10 anos e proveniência da Região Sudeste do Brasil. Alterações hematológicas, dor óssea, hepatomegalia, esplenomegalia, cansaço foram os sinais e sintomas mais recorrentes. Os diagnósticos inferidos para a população estudada foram: Risco de sangramento; Fadiga; Dor crônica e Dor aguda; Mobilidade física prejudicada; Nutrição desequilibrada: menos do que as necessidades corporais; e Risco de Desenvolvimento atrasado. **Conclusão**: O estabelecimento dos Diagnósticos de Enfermagem prioritários a partir dos sinais e sintomas possibilita alcançar resultados esperados a cada indivíduo no contexto do cuidado.

Descritores: Doença de Gaucher; Síndrome de Liposidose por Cerebrosídeos; Sinais e Sintomas; Processo de Enfermagem; Diagnóstico de Enfermagem.

RESUMEN

Objetivo: Identificar las señales y los síntomas de pacientes con Enfermedad de Gaucher, infiriendo los posibles diagnósticos prioritarios de enfermería. **Método:** Estudio transversal, desarrollado entre 2013 y 2015 en un laboratorio especializado. La muestra (n=91) estaba constituida por los registros de pacientes con diagnóstico genético de la Enfermedad de Gaucher. El estudio respetó las normas de la investigación. **Resultados**: Prevaleció el sexo femenino (57,1%), con franja de edad entre 0 y 10 años y procedencia de la Región Sureste de Brasil. Las alteraciones hematológicas, el dolor óseo, la hepatomegalia, la esplenomegalia y el cansancio fueron las señales y los síntomas más recurrentes. Los diagnósticos inferidos de la población estudiada fueron: Riesgo de sangrado; Fatiga; Dolor crónico y Dolor agudo; Movilidad física perjudicada; Nutrición desequilibrada: menos que las necesidades corporales; y Riesgo de Desarrollo

atrasado. **Conclusión**: El establecimiento de los diagnósticos prioritarios de enfermería, a partir de las señales y de los síntomas, permite obtener los resultados esperados para cada individuo en el contexto del cuidado.
Descriptores: Enfermedad de Gaucher; Síndrome de Lipidosis por Cerebrósidos; Señales y Síntomas; Proceso de Enfermería; Diagnóstico de Enfermería.

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INTRODUCTION

Gaucher disease is an inborn error of metabolism in the lysosomal storage diseases group, and is the most frequent within this group, with a prevalence of approximately 1:70,000 live births. It is of autosomal recessive inheritance, caused by deficiency of the acidic beta-glucosidase enzyme, which leads to the accumulation of glycosylceramide, a glucocerebroside, in tissue macrophages. This compromises the spleen, liver, bone marrow, central nervous system, lung and lymph nodes and can contribute to pancytopenia and massive hepatosplenomegaly[1-2].

Patients may have a long asymptomatic course, with few signs of disease until adulthood, or more frequently manifest clinical symptoms as early as the first years of life[1]. The clinical or phenotypic manifestations of Gaucher disease will depend on the degree of acidic beta-glycosidase deficiency and the accumulation of glycolipids, which, because of their variability, will differentiate the clinical types of the disease: type 1, type 2 and type 3. In types 2 and 3, there is neurological impairment, while in type 1 there is a greater presence of signs and symptoms, as well as a higher prevalence in the affected population[3].

Gaucher disease is not a very frequent condition in the general population, and its definitive diagnosis is difficult, because it presents a similar picture to that of diseases with high prevalence. An early and specific clinical diagnosis is needed, together with a multidisciplinary follow-up, for a good clinical resolution[3].

Information on Gaucher disease in the academic and clinical setting, as well as in nursing practice, is little publicized. Thus, a characterization of the signs and symptoms of the disease is relevant to enable an early clinical and nursing diagnosis that favors a care plan for the affected population.

The set of signs and symptoms identified from the clinical evaluation is the defining characteristic that guides the generation of diagnostic hypotheses and help in the selection of a particular Priority Nursing Diagnosis. For NANDA-I[4-5], Nursing Diagnosis is defined as the clinical judgment about a human response to health conditions/life process, or a vulnerability to that response, by an individual, family, group, or community.

Thus, the basis for the elaboration of a Nursing Diagnosis is a broad data collection focused on the needs (or problems) of the individual. The diagnosis stage is inserted in the Nursing Process, and has five interrelated steps[6] that subsidize and assist nurses' understanding of observed situations, making judgments and executing necessary actions, in order to achieve an outcome, therefore it is of great importance in clinical practice.

Thus, based on the signs and symptoms that lead to the defining characteristics and, consequently, to the related factors, it is possible to identify Nursing Diagnoses that will subsidize more individualized care for patients with Gaucher disease.

In this perspective, the present study aims to identify the signs and symptoms of patients with Gaucher disease, in order to infer the possible Priority Nursing Diagnoses for these patients.

METHOD

Ethical aspects

The study was approved by the Research Ethics Committee of the Federal University of Rio Grande do Sul (UFRGS), and all ethical and legal research norms were complied with, according to Resolution 466/12 of the National Health Council[7].

Study design, location and period

A cross-sectional study, conducted at the Laboratory of Inborn Errors of Metabolism - Lysosomal Storage Diseases of the Department of Biochemistry, Institute of Basic Health Sciences of UFRGS, from January 2013 to December 2015.

Population or sample; inclusion or exclusion criteria

The study population consisted of the records of 150 patients with suspected Gaucher disease, registered in a database. These patients came from different geographic regions of Brazil, and their data, referring to Gaucher disease, were sent from a reference hospital for the patient to the Laboratory of Inborn Errors of Metabolism, with the purpose of a specialized study.

All patients with confirmed diagnosis of Gaucher disease were included in the study. Such that 59 patients were excluded from the total sample, because their diagnosis had not been confirmed. Therefore, the sample comprised 91 patients with genetic confirmation for Gaucher disease, after laboratory tests and clinical evaluation.

Study protocol

The clinical reasoning and diagnostic judgment to infer the Nursing Diagnoses from the identified signs and symptoms relied on the clinical experience of the researchers, as well as on their knowledge of the standardized NANDA-I language[5].

Analysis of results and statistics

The variables analyzed were: age, sex, signs and symptoms related to the underlying pathology, consanguinity, occurrence of familial cases and geographic region of Brazil where the reference hospital for the patient is located. The results were expressed as mean, standard deviation of the mean and relative frequency, using Statistical Package for the Social Sciences (SPSS) version 18.0.

RESULTS

The sample consisted of records from 91 Gaucher Disease patients, with a mean age of 26 (SD = 18.4) years, at the time they were entered into the database.

Of the total sample, there was a predominance of females (57.1%); age at diagnosis between 0 and 10 years of age (26.37%); from the Southeast Region (41.7%); and referred from urban areas (71.4%). The sample demographics are described in Table 1.

Table 1 – Demographic characterization of patients with Gaucher disease, Porto Alegre, Rio Grande do Sul, Brazil, 2013-2015

	n (N = 91)	%
Sex		
Female	52	57.1
Male	39	42.9
Age group (years)		
0 ⊢ 10	24	26.4
11 ⊢ 20	19	20.9
21 ⊢ 30	16	17.6
31 ⊢ 40	12	13.2
41 ⊢ 50	06	6.6
51 ⊢ 60	11	12.0
61 ⊢ 70	03	3.3
Demographic region (Brazil)		
Southeast	38	41.7
Northeast	31	34.1
South	08	8.8
Midwest	08	8.8
North	06	6.6
Location of hospital (zone)		
Urban	65	71.4
Rural	26	28.6

Note: n = sample; % = relative frequency.

Considering Gaucher disease is of genetic origin, the diagnosis was confirmed in the same line of hereditary succession in 15.4% of the cases and in 13.2% by family screening. Two (2.2%) cases were found with a history of consanguinity between the parents.

Regarding the clinical profile of the patients, the main signs identified were: splenomegaly, hematological alterations (anemia and thrombocytopenia), hepatomegaly and bone pain. It is underscored that most of the patients presented at least one clinical sign (45.1%), followed by patients who presented two (34.1%), and three or more (4.4%) signs. The majority of patients presented at least two symptoms (36.3%), followed by patients presenting at least one (17.6%), three (16.5%) and four or more (4.4%) symptoms.

The most frequently identified symptoms were: fatigue; bone and joint pain; lack of appetite; and abdominal discomfort.

Data referring to the main signs and symptoms recorded are described in Table 2.

According to the symptomatology and the three clinical types of Gaucher disease, there was a predominance of type 1 (79.1%) in relation to type 2 (7.7%) and type 3 (13.2%).

Table 2 – Principal signs and symptoms of patients with Gaucher disease, Porto Alegre, Rio Grande do Sul, Brazil, 2013-2015

	n	%
Signs		
Splenomegaly	45	49.5
Hematologic Changes	36	39.6
Hepatomegaly	33	36.3
Bone pain	28	30.8
Weight Loss	27	29.7
Epilepsy	10	11.0
Mental retardation	8	8.8
Hypertonia	8	8.8
Symptoms		
Fatigue	24	26.4
Joint pain	24	26.4
Loss of appetite	22	24.2
Abdominal discomfort	21	23.1
Abdominal pain	15	16.5
Difficulty with movements	14	15.4
Increased active bleeding in tissue injury	7	7.7

From the identification of the signs and symptoms and determining the defining characteristics, it was possible to match these characteristics to the related factors and then to list the priority Nursing Diagnostics for the studied population: Chronic pain; Acute pain; Fatigue; Risk for bleeding; Impaired physical mobility; Imbalanced nutrition: less than body requirements; and Risk for developmental delay. Data on the priority nursing diagnoses are described in Chart 1.

Chart 1 – Signs and symptoms, defining characteristics, and related factors to the Priority Nursing Diagnoses identified, Porto Alegre, Rio Grande do Sul, Brazil, 2016

Signs	Symptoms	Defining characteristics / Risk factors *	Related factor *	Nursing Diagnoses (number)*
Bone pain	Pain in bones and joints for more than three months	Change in ability to continue previous activities; Verbal report of pain.	Genetic disorder	Chronic pain (00133)

To be continued

Chart 1 (concluded)

Signs	Symptoms	Defining characteristics / Risk factors *	Related factor *	Nursing Diagnoses (number)*
Hematologic Changes (Anemia and thrombocytopenia)	Increased active bleeding in tissue injury	Inherent coagulopathies; Impaired hepatic function; Abnormal blood profiles.	–	Risk for bleeding (00206)
_	Fatigue	Lack of energy; Report of tiredness.	Anemia	Fatigue (00093)
Hypertonia	Difficulty with movement	Limited ability to perform fine and gross motor skills; Postural instability; Response time decreased.	Contractures; Decreased muscle control; Discomfort; Pain; Joint stiffness.	Impaired physical mobility (00085)
Esophageal Varices Hepatomegaly	Pain and abdominal discomfort	Changes in appetite; Self-report of pain characteristics using a standardized pain instrument.	_	Acute Pain (00132)
Splenomegaly Weight Loss	Loss of appetite	Abdominal cramps; Abdominal pain; Satiety immediately after eating food.	Insufficient food intake	Imbalanced nutrition: less than body requirements (000002)
Mental retardation / Epilepsy	–	Convulsions Genetic disorder Inadequate nutrition	–	Risk for developmental delay (00112)

Source: NANDA, North American Nursing Diagnosis Association: definitions and classification*[5].**

DISCUSSION

Gaucher disease is inherited in an autosomal recessive pattern and can affect both sexes, with no female or male sex bias; however, in the present study, there was a predominance of females. This data corroborates a study that evaluated the impact of enzyme therapy for patients with Gaucher disease in clinical and laboratory parameters after two, five and ten years of treatment, where 61.5% of the sample was female[8]. Similarly, the Gaucher Register of the International Collaborative Gaucher Group (ICGG) – a multicenter registry with demographic and clinical results, created in 1991 – showed that in Brazil, out of a total of 551 registered and accompanied patients, 58% were female, closely matching world data (53%)[9]. Data showing the predominance of females with Gaucher disease can be attributed to the fact that women are more interested in health issues and therefore have regular consultations, which favors the possibility of a clinical diagnosis.

As to age, the present study shows that the highest number of patients diagnosed with Gaucher disease is in the age group between 0 and 10 years of age, although the average age is 26 (SD = 18.4) years. Confirmation of the diagnosis in earlier age groups is in agreement with data found in a study that described the clinical and laboratory characteristics of children with Gaucher disease and exacerbated symptoms with evolution to enzyme replacement therapy[2]. Similar values are also reported in a study conducted in 2016, where the mean age of the patients was 26 years (range 5 to 60 years) [10]. Thus, the diagnosis established at the onset of childhood may be elicited by greater severity of the disease, a period in which the signs and symptoms are accentuated, or by regular follow-up in consultation with the pediatrician. Irrespective of the conditions of how the diagnosis was established, the precocity of diagnosis favors good disease control and provides a positive impact on patients' quality of life.

Gaucher disease presents in both the lethal form in the perinatal phase and an asymptomatic form. The literature describes three clinical types for Gaucher disease, based on absence (type 1) or presence and severity of neurological impairment (types 2 and 3). Type 1 is the most common form.

According to the ICGG[11], there are approximately 5,000 individuals diagnosed with Type 1 Gaucher disease in the world. Of these, approximately 500 patients are in Brazil, ranking the third country with the highest number of patients identified, after the United States (45%) and Israel (17%). In November 2010, the number of patients registered in the ICGG reached 5,914, of which 561 were in Brazil[12]. Currently, with the increase in density of the Brazilian population, the diagnosis of Gaucher disease could exceed 1,000 cases.

The Brazilian population is distributed irregularly, with large concentrations in urban centers. The majority of cases identified in this study come from Southeast Brazil, where the population is more numerous and where there are specialized reference centers for the establishment of clinical diagnosis. According to Pires and Sobreira (2003), if we consider the population density of Brazil and the regional distribution of Gaucher disease, we can see the existence of a relationship between these two parameters[13]. The National Association of Patients with Gaucher Disease reported that in May 2011, there was a predominance of patients in the Southeast Region (57%), with prevalence of patients in São Paulo, followed by Minas Gerais and Rio de Janeiro[14]. The same data was evidenced in the present study, in which the Southeastern Region and the state of São Paulo had the highest percentages of confirmation of the biochemical diagnosis.

The diagnosis of an affected person leads to the need for family assessment. At first, this assessment involves molecular and enzymatic testing of parents and others at risk of being carriers or affected. Since patients with type 1 Gaucher disease may be asymptomatic or symptomatic as yet undiagnosed, it is important to counsel families that the test may reveal other affected persons and not just carriers. This may be advantageous in symptomatic undiagnosed patients, because they would benefit from correct diagnosis and potential treatment[15]. In the present study, patients were diagnosed with Gaucher Disease by family screening (13.2%) and family cases (15.4%).

Regarding the history of consanguinity, in a study that aimed to identify clinical and laboratory characteristics of children with Gaucher disease, a history of consanguinity was present in 93.3%[2]. In the present sample, in 2.5% of the patients, consanguinity among the parents was identified.

Gaucher disease encompasses a set of differentiated signs and symptoms according to types 1, 2 and 3.

Type 1 – clinical manifestations may occur from infancy to late in adult life. Among the signs presented by type 1 patients, splenomegaly and hepatomegaly stand out as the most common clinical features observed[16]. In a study cited above[2] all patients presented with splenomegaly, while hepatomegaly was identified in 86.7% of the sample. In the present study, patients with splenomegaly (49.5%) and hepatomegaly (36.3%) predominated in an age group ranging from 0 to 65 years, with a consequent clinical picture for esophageal varices (5.5%).

Bone involvement is responsible for much of the morbidity presented by patients with Gaucher disease. Bone and joint pain, often associated with the pain crisis, can be debilitating and chronic. Radiographic findings indicate a certain degree of bone marrow infiltration and replacement by Gaucher cells, resulting in loss of bone trabeculation and decreased density, more common in the epiphysis and metaphysis of long bones[17]. A study carried out in 2012, aimed to highlight Gaucher disease, in order to improve patient care and quality of life, showed that in 80% of the sample there was confirmation of bone disease by radiographic and historical examination of bone pain[18]. In addition, a study[2] mentioned above, reported approximately 50% of the patients presented with bone pain. Likewise, approximately one-third of the patients in the current sample presented bone pain as a sign and pain in bones and joints as a symptom, and 15.4% also reported difficulties with movement.

Hematological changes occur as a consequence of sequestration and splenic hyperactivity and of spinal cord infiltration by Gaucher cells. The most frequent findings are thrombocytopenia and anemia[19], corroborating our findings, of 39.6% patients with anemia and thrombocytopenia. These data are also comparable to those found in the ICGG[9], in which the frequency of hematological changes reaches up to two thirds of the patients. In addition to a low platelet count, a clotting factor deficiency has also been described, which may contribute to the tendency for bleeding[19]. Patients with Gaucher disease, in addition to depicting hematological disorders, bone and visceral manifestations, may present chronic fatigue as a consequence of hematological disorders[20]. Data from the present study are similar to those previously found, in which approximately 26.4% of the sample reported fatigue and/or lack of energy and 7.7% presented increased active bleeding in tissue injury.

Type 2 – The acute neuropathic form, affects infants at 4-5 months of age, compromising brain, spleen, liver and lungs. The neurological picture is severe, with multiple seizures, hypertonia, apnea and progressive mental retardation. The evolution is rapid, with death in the first two years of life, usually due to pulmonary involvement[17-21].

Type 3 – Patients with type 3 (chronic neuropathic form) are mostly children and adolescents, with impairment of brain, spleen, liver and bones. Clinical manifestations include growth retardation, hypersplenism, and skeletal changes. The evolution of the neurological picture is variable, but less severe than that of type 2. Survival occurs until the second or third decade of life[17-21].

Considering the sample studied, the most prevalent signs and symptoms were: epilepsy (10.98%), mental retardation (8.79%) and hypertonia (8.79%), thereby identifying patients with types 2 and 3.

The signs and symptoms presented by patients are the indicators, that is, the defining characteristics, and the clinical evidence of the patient specifically with Gaucher disease. Thus, using clinical judgment, it is possible to identify more accurately the possible Nursing Diagnoses for the sample studied.

Of the defining characteristics described in NANDA-I[5], the following were identified in the patients: verbal complaint of pain and altered ability to continue previous activities, both of which are listed as factors related to Genetic Disorder. These findings confer with the aforementioned[22-24] on high morbidity in patients with Gaucher disease, in the existence of a frequent concomitance of pain with limitations of mobility and performance of activities. These defining characteristics, when combined, have an extremely negative impact on patients' quality of life. Therefore, the priority Nursing Diagnosis identified for the defining characteristics and related factor was Chronic Pain, defined as:

> Unpleasant sensory and emotional experience associated with actual or potential tissue injury, or described in terms of such injury; sudden or slow onset of any light to intense, constant or recurrent intensity without an anticipated or predictable termination and lasting more than three months[5].

Bone manifestations in affected patients are caused by several factors and may include bone marrow infiltration, severe bone crises, intermittent chronic bone pain, osteopenia, osteoporosis, and pathological fractures of long bones and vertebrae. The present study identified the priority Nursing Diagnosis as "Impaired Physical Mobility", which contemplates two observed signs, hypertonia and bone pain, and is defined as "limitation of independent and voluntary physical movement of the body or of one or more extremities"[5].

The Nursing Diagnosis "Impaired Physical Mobility" was substantiated with the following defining characteristics: limited ability to perform fine and gross motor skills, postural instability, decreased response time, all related to decreased muscle strength, decreased muscular control, decreased resistance, contractures, discomfort, pain and joint stiffness.

The "tiredness" symptom predominated in 24% of the studied sample and is a defining feature of the "Fatigue" Nursing Diagnosis, which is conceptualized as "an oppressive and prolonged feeling of exhaustion and diminished ability to perform physical and mental work at the usual level"[5]. Patients with Gaucher disease may suffer from chronic fatigue, which causes functional disability and negatively affects their quality of life; manifestations of the disease, such as anemia and bone pain, can cause or contribute to fatigue[25].

In a study of 218 patients with Gaucher disease, four symptoms were related with hepatosplenomegaly: abdominal pain; precocious satiety; abdominal discomfort; and acute colic in the upper quadrant[26]. These data are similar to the present study, where the following defining characteristics were identified in the patients with hepatosplenomegaly: changes in appetite, verbal report of pain, abdominal cramps, abdominal pain and satiety immediately after ingestion, all related to an inability to digest the foods. These defining characteristics and related factors made it possible to prioritize the Nursing Diagnoses "Acute Pain" and "Imbalanced Nutrition: less than body requirements". For the Nursing Diagnosis "Acute Pain', is defined as "unpleasant sensory and emotional experience associated with actual or potential tissue injury, or described in terms of such injury; Sudden or slow onset, of mild to severe intensity, with an anticipated or predictable termination"[5]. For the Nursing Diagnosis "Imbalanced nutrition: less than body requirements", we find as a definition "Insufficient intake of nutrients to satisfy the metabolic requirements"[5].

Risk diagnostics does not consider signs and symptoms, but risk factors. As such, the possible Nursing Diagnosis referring to hematological alterations is "Risk for Bleeding", defined as "vulnerability to reduction in blood volume that can compromise health"[5]. This priority diagnosis presents inherent coagulopathies and impaired hepatic function as risk factors, evidenced in the present study by anemia, thrombocytopenia and hepatomegaly.

For the signs "mental retardation" and "epilepsy", the Nursing Diagnosis "Risk for Developmental Delay" was defined as "vulnerability to delay of 25% or more in one or more areas of social or self-regulatory behavior, or in cognitive, language and gross or fine motor skills, which may compromise health"[5]. This diagnosis presents as risk factors: convulsions and genetic disorders.

The above mentioned Nursing Diagnoses are in agreement with the defining characteristics and related factors selected from the clinical complaints of patients with Gaucher disease, which provides us a reliable clinical judgment in the choice of interventions and nursing actions that qualify the care.

Study limitations

A limitation was the absence of complementary data in patients' records, stored in a database; If these were available, they could illustrate other relevant results for nursing practice.

Contributions to Nursing

The study could favor future nursing actions that meet the needs of patients with Gaucher Disease, as well as to improve care protocols and thus contribute to individualized care of these patients.

CONCLUSION

The Nursing Diagnoses "Fatigue", "Risk for Bleeding", "Chronic Pain", "Acute Pain", "Impaired Physical Mobility", "Imbalanced Nutrition: less than Body Requirements", and "Risk for Developmental Delay" were inferred from the signs and symptoms identified in the sample studied, this reinforces the selection of these Nursing Diagnostics as a priority in the care of patients with Gaucher Disease.

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