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Nursing care for children with Pompe disease: a case study

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ABSTRACT

Aim: to identify diagnoses, interventions and expected nursing outcomes for the child with Pompe disease using the International Classification for Nursing Practice® 2017. **Method:** case study conducted in December 2017, with a mother of child with Pompe disease. **Results:** seventeen nursing diagnoses were presented, distributed among the demands of medicated, technological, habitual, modified, developmental, and family care. **Discussion:** the nursing diagnoses were related to the treatment and complications presented by the child as a result of Pompe disease, increased by the family needs to develop the care at home. **Conclusion**: it was verified that the classification of the nursing practices allowed creating more specialized possibilities to elaborate and execute a care plan based on the real needs of this child, allowing the possibility of generalization for the patients with the same disease.

Descriptors: Glycogen Storage Disease Type II; Pediatric Nursing; Primary Health Care; Nursing diagnosis; Family.

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INTRODUCTION

Pompe disease or type 2 glycogenosis is a rare, progressive muscle disease triggered by an autosomal recessive disorder with alpha-1,4-glycosidase (GAA) enzyme deficiency, responsible for the breakdown of glycogen in the cell lysosome, generating a broad clinical spectrum[1-2].

The incidence varies according to ethnicity and geographic region, estimating, worldwide, one case per 40,000 live births, and in Brazil, it is believed that there are about 120 patients[3-4].

The clinical manifestations of Pompe disease are heterogeneous, but similar to numerous other diseases, which can make its diagnosis difficult and time-consuming. Among the main manifestations of the disease are: hypotonia, delayed motor development, muscular weakness, swallowing problems, and heart and respiratory failure[3-4].

Considering the difficulty in the diagnosis of rare diseases, people with Pompe disease may spend months or even years using numerous health services, and are submitted to inadequate interventions until reaching their definitive diagnosis and initiating the effective treatment[3-4].

One of the available treatments is enzyme replacement therapy, which uses recombinant GAA (Myozyme®)[5] administered in a hospital environment and by a specialized health team.

For the health care of people with rare diseases, such as Pompe disease, the National Policy for Comprehensive Care for Persons with Rare Diseases was instituted in Brazil in 2014, with the objective of contributing to the reduction of morbidity and mortality of secondary manifestations. Among its guidelines, this policy seeks to improve people's quality of life by means of actions of promotion, prevention, early detection, timely treatment, disability reduction and palliative care[6].

Thus, although the priority treatment for Pompe's disease happens in the hospital, the care for his health should be continuous, systematized and specific, shared among families and primary health care teams. Therefore, the needs demanded by these patients and their families should be recognized by the multiprofessional team, so as to offer them the conditions to enhance this care in the home environment and improve the quality of life.

In this perspective, considering the nurse part of this team, it should be able to fully serve this clientele, using the nursing process as a tool to order, systematize and execute their practices according to the demands presented[7].

Based on these premises and seeking to know scientific evidences for the integral care of this clientele, this study aims to identify diagnoses, interventions and expected results of nursing to the children with Pompe disease using the International Classification for Nursing Practice (CIPE, acronym in Portuguese) 2017.

METHOD

This is a case study on a child with Pompe disease assisted in a hospital located in the southern region of Brazil. The case study is a type of qualitative approach research for complex investigations, and can be developed with a single case[8], as the rare disease referred to here.

The data search was performed in the electronic medical record and through a semi-structured interview with the mother, during one of the hospitalizations of the child in said

hospital for enzyme replacement therapy in December 2017.

A pre-elaborated road map containing birth data and the child's special health needs (presented in the case description) was used; maternal data (age, marital status, schooling) and socio-demographic data of the family (number of people in the household, family income, government benefits received, type of housing).

For conducting the interview, the genogram and ecomap instruments were used as an approximation strategy, and then the following guiding question was asked: "Tell me about your experience in child care since the emergence of the special need of health". It is emphasized that this interview was performed by a nurse, in a private setting, after the consent of the mother, and was recorded in audio and transcribed in full.

From the interview it was possible to describe the trajectory traversed by the family in search of attention to the child's health, as well as to identify the demands of care, classified as: medicamentous, technological, habitual and developmental[9]. Considering the needs that the family presented in this trajectory, the authors added the demand for family care.

Based on the demands of care presented by the child and his/her family, CIPE® was used to identify the nursing diagnoses and interventions, seeking the integral care of the child with Pompe disease.

The authors' preference to take care of the demands for care to identify nursing diagnoses, in this case, is due to the complexity of the specialized and long-term care required by the child[9].

With respect to the detailing of the rationale of the diagnoses, a conceptual map was first constructed with the objective of demonstrating the relationship and the grouping of care needs with the concepts that form the diagnoses. In this thinking, we considered the training concepts: signs and symptoms, risk factors and living conditions, which were organized following the classification of care demands[7].

Then, as recommended by CIPE®, the nursing diagnosis statement was constructed using a term from the **Focus** axis and a term from the **Judgment** axis, plus additional terms when necessary; and also to determine these two elements in nursing practice, the term nursing diagnosis was used[10].

After constructing the diagnostic statements, the nursing interventions were elaborated using a term for the **Action** axis and a term for the **Target** axis, adding additional terms as needed. From this moment on, it is possible to list the priority nursing interventions to attend to the demands of care presented[10].

This study was approved by the Research Ethics Committee of the University of São Paulo at Ribeirão Preto College of Nursing, under the no. 2,000,924 report on April 5, 2017, pursuant to Resolution 466/2012.

RESULTS

This session describes the case of a child with Pompe disease in order to identify their needs and demands for care, as well as their families, for the subsequent construction of nursing diagnoses and interventions, based on CIPE® 2017.

Description of the case

Female child, henceforth identified as Flor, who at the time of the interview was 2

years and 9 months old. She lives with her father (26 years old) and her mother (24 years old) in a masonry house provided by the town hall.

She was born in a surgical delivery, with a gestational age of 37 weeks, weighing 2,925 g, with Apgar 6 in the 1st and 9 in the 5th minute. She was born at the usual risk hospital in her city and because she presented important hypotonia and cyanosis she was referred to a high risk service in the neighboring municipality, but only at eight months did she go to the neuropediatrician. He suspected Pompe's illness and informed his parents that the child would live up to one year and three months. A GAA test was performed to confirm the diagnosis. When Flor was nine months old, they received confirmation of the diagnosis. The following month, enzyme replacement therapy (Myozyme GENZYME®) was started, performed by means of venous infusion every two weeks in a hospital setting.

Two months after starting treatment, Flor presented a significant clinical evolution, was able to crawl, sit and take the first steps. However, at one year and three months she presented pneumonia, because, according to her mother, she had recurrent bronchoaspirations. It was a period of seven months of hospitalization, which impacted the routine, finances and emotions of the family, considering that they reside in another municipality. During this period, Flor presented cardiorespiratory arrest during enzymatic therapy, causing greater impairment to the neuropsychomotor system. She was discharged with a tracheostomy, using mechanical ventilation, nasoenteral probe for diet and medication, and port-a-cath for the administration of enzyme therapy.

After this hospitalization, her parents dedicate themselves entirely to Flor's care. Tracheostomy and airway aspiration care, medication and feeding administration and first aid techniques were learned in the hospital. The mother said she feels empowered for day care and has her husband's source of support in home care. The family has the hospital, where she undergo enzymatic therapy, as a reference and considers it as their second home.

Although they receive financial aid and disposable materials from the municipal health department, the mother wants more involvement of the primary health care team (PHC) of her municipality in home care.

Conceptual map: elaboration of the diagnoses

The case study presented identified the needs and demands of care: medical, technological, customary, development and family care. Based on these demands, 17 nursing diagnoses were shown, which can be visualized in Figure 1.

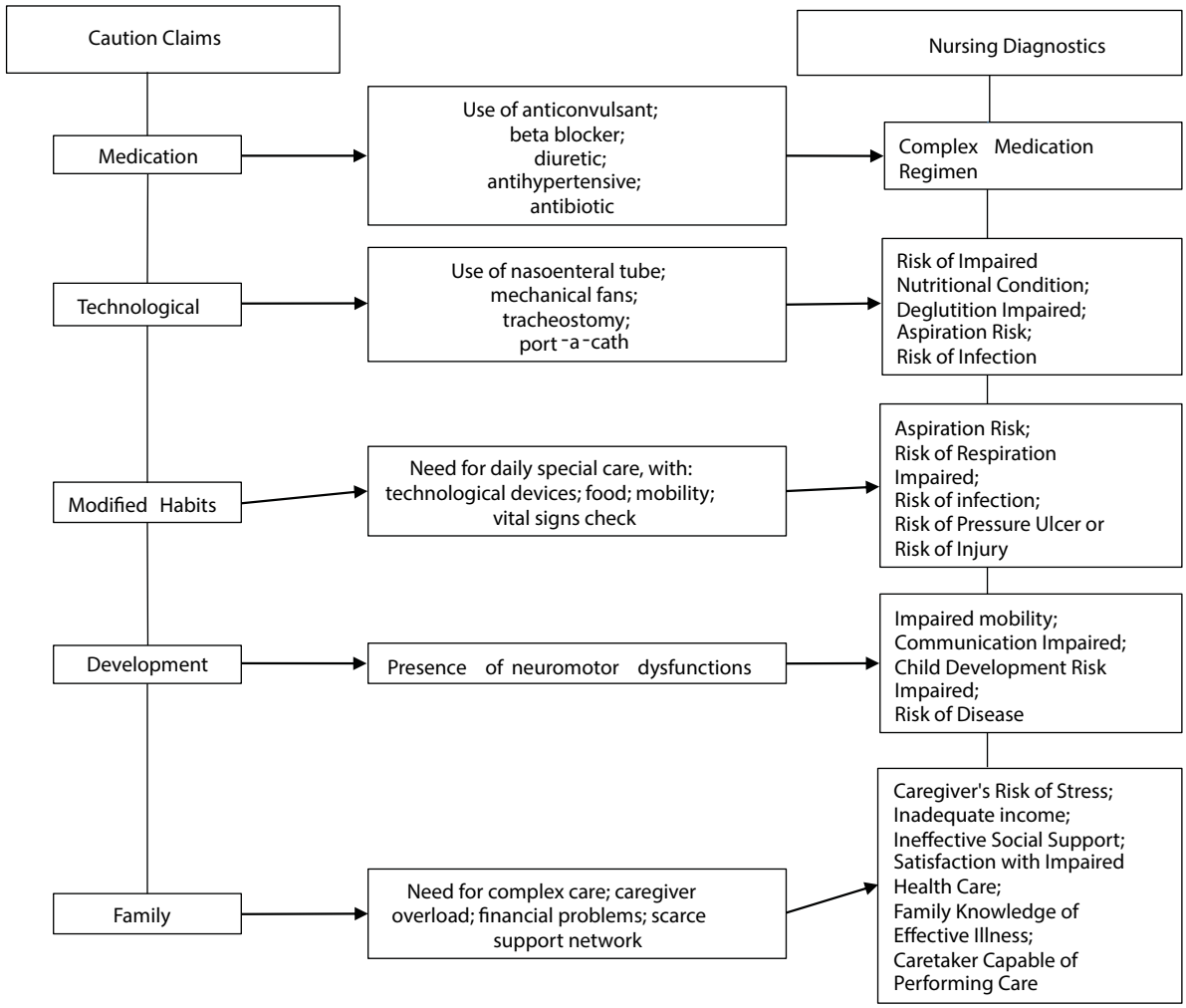
In order to meet the needs of the child and the family in promoting care for PHC services, Figure 2 presents the diagnoses with their respective nursing interventions proposed for comprehensive care of children with Pompe disease and their family.

DISCUSSION

The child with Pompe disease has peculiar health needs that require hospital, home and PHC services. In this way, it represents a great challenge for families and professionals, considering the need for continuous care and complex nature.

In the present case study, the relationship between the hospital care team and the

Figure 1. Nursing diagnoses in a child with Pompe disease, according to the identified care demands. South Region, Brazil, 2017



Source: research data.

family became close because of their proximity to care, hospitalization time, and biweekly frequency for the administration of enzyme therapy and mutual trust that was established.

However, only the hospital bond was insufficient to provide full care to children with Pompe disease, demonstrating the need to strengthen ties with PHC professionals, so that the family could receive support to meet the demands for care presented and establish habits to enhance the child's living conditions[6].

Therefore, the proposal of a care plan directed to the PHC services for this scenario became fundamental to give visibility to the needs in the domicile scope, since the care did not end in the hospital discharge. In this perspective, designing a care plan, based on the nursing process, is a guideline so that the care provided to the child becomes effective[9].

The use of the nursing process can benefit patients by ensuring individualized care, improving the relationship between professionals, patients and caregivers, resulting in

Figure 2. Diagnoses, interventions and expected nursing outcomes according to the care demands of children with Pompe disease and their family. South Region, Brazil, 2017

Nursing Diagnostics	Nursing Interventions	Expected Nursing Outcomes
Complex Medication Regimen	Guide and demonstrate care in handling, dosing, dilution and administration via probe; Guidance on therapeutic regimen and possible side effects; Reinforce maintenance of treatment; Schedule home visits for supervision.	Adherence to the drug regimen
Risk of Impaired Nutritional Condition	Guidance on enteral nutrition; Monitor intake, flow and weight; Request follow-up nutritionist to advise on the diet; Monitor growth and development through surveillance tools advocated by the Ministry of Health.	Reduced risk of impaired nutritional status; Improved nutritional status
Impaired Deglutition	Refer the speech pathologist to improve swallowing reflexes.	Improved swallowing; Adequate swallowing
Aspiration Risk	Guide bed head elevation to 90 degrees during meals and maintain position for 30 minutes; Observe and suspend your diet if you experience cyanosis, dyspnoea or asphyxia during infusion; Wash the catheter with water after administration of the diet; Observe and report the presence of abdominal distension.	Decreased aspiration risk; Risk of aspiration absent
Risk of Infection	Direct signs/symptoms of infection (local heat, redness, bleeding, pain and edema) at the site of insertion of the port-a-cath and tracheostomy and report changes to the health team; Orientation of the family on infection prevention (hygiene of the hands before caring for the child, use of sterile materials in the aspiration of the tracheostomy and airways, exchange and keep clean and dry dressings).	Decreased risk of infection; Risk of infection absent
Risk of Impaired Breathing	Direct vital signs monitoring (check of respiratory movements, blood pressure, radial pulse, body temperature); Keep headboard high for easy breathing; Make a change of decubitus frequently; Guidance for aspiration of the tracheostomy and airways whenever necessary; Guidance on mechanical ventilator and tracheostomy care; Request and refer to respiratory physiotherapy, including preventive.	Decreased risk of impaired breathing; Risk of impaired respiration absent
Risk of Pressure Ulcer or Risk of Injury	Guide the performance of the change of decubitus and passive movements to reduce pressure and avoid injuries; Use preventive skin protection devices, such as foam mattresses, pressure-alternating mattresses, cushions or pillows to avoid discomfort; Implement a daily routine of inspection and skin care; The health team will be able to use the Braden Q (pediatric) scale to assess the Risk of Pressure Ulcers.	Decreased risk of pressure ulcer or injury; Risk of pressure ulcer or absent injury;
Impaired mobility	Guide passive movements with the child every two hours; Monitor and record daily any signs of immobility complications (contractures, venous stasis, thrombosis, pneumonia, urinary tract infection); Guide family members about bed hygiene (clean, waterproof bedding, pillows for protection and comfort); Request and refer for motor physiotherapy to develop a mobility recovery plan.	Improved mobility; Adequate mobility

Communication Impaired	Guide the caregiver to create opportunities and communication activities for the child according to age; Reinforce to the mother or caregiver the stimulus to speech with the child; Refer to speech therapist to improve stimuli / verbal communication; Guide to maintaining a calm and non--threatening environment by reducing excessive environmental stimuli.	Improved communication; Effective communication
Child Development Risk Impaired	Guide parents about age-related developmental milestones; Stimulation with toys of the age; Investigate the level of development of the child in all areas of functioning; Praise the mother or caregiver to continue to stimulate the child and the importance of this process.	Reduced risk of impaired child development; Adequate child development
Risk of Disease	To guide the caregiver as to the importance of the vaccines, amelioration, application technique, effects; Update the vaccination schedule, including influenza and pneumococcus; Assess immunization status.	Absence of disease risk; Decreased risk of disease
Caregiver's Risk of Stress	Support the caregiver and provide guidelines to feel safe in care; Support family coping process; Forward to psychological care.	Stress risk of absent caregiver; Decreased risk of caregiver stress
Inadequate Income	Guidance on the rights of children with special health needs (explain patients' rights); Request social service support.	Improved Income; Adequate income
Ineffective Social Support	Guide to find a family support network to assist in activities; Support the family to identify their own resources, community and social projects to face daily difficulties; Encourage relationships with people with common interests and goals; Identify friends, neighbors or family members who can support the family; Promote social support; Assist the family to identify and collaborate for their integration in support groups for people with rare diseases.	Improved Social Support; Effective Social Support
Satisfaction with Impaired Health Care	Carry out monitoring and guidance effectively; Schedule home monitoring; Provide health promotion service; Establish a link between Health Unit, social teams and family; Welcoming the family in their needs; Assess satisfaction with health care.	Satisfaction with improved health care
Family Knowledge of Effective Illness	Forward to genetic counseling and family planning; Conduct guidelines and conduct active search among siblings, when available; Assess the level of family knowledge about the disease.	Family Knowledge About Effective Illness
Caretaker Capable of Performing Care	Praise the mother or caregiver; Provide caregiver support for disease management.	Caregiver able to perform care

Fonte: dados da pesquisa.

qualified and humanized care. For the nursing team, its use will focus on professional qualification and, consequently, valorization, recognition, legal support and optimization of care and research[11].

The present care plan included diagnoses, interventions and expected nursing outcomes. The elaboration of these diagnoses from CIPE® sought to improve the quality of care by the PHC, with a view to enhancing home care. Through this classification, nurses construct the statements of the diagnoses by the clinical presentation of the people who are in their care, since the terms occur in their

practice and require the clinical reasoning of the problems[12].

In this study, it was verified that the majority of nursing diagnoses were related to the complications resulting from the progression of Pompe disease, plus complications resulting from prolonged hospitalization. It should be emphasized that this disease causes glycogen accumulation in the tissues, causing difficulties in breathing, locomotion, speech and hearing[5,13], causing patients to require technological support to maintain their vital functions.

The diagnosis of complex drug regimen, one of the 17 listed in this study, addressed the demand for medication care, considering the children's need to make use of an amplitude of medications, which potentiate their clinical recovery related to the disease itself and the iatrogenies of the hospitalization period.

Four nursing diagnoses were directly related to the presence of technological devices: nasoenteral catheter, mechanical ventilator, tracheostomy and port-a-cath. Consequently, other diagnoses were included as a demand for usual care; however, its implementation refers to the use and maintenance of these devices by the family. This reveals the complexity of technology-dependent child care, corroborating another study that pointed out the need for family members and caregivers to acquire new skills and competencies for care[14].

The diagnoses Risk of Impaired Nutritional Condition and Impaired Deglutition relate to the feeding of the child by nasoenteral tube. The use of the probe is due to the presence of muscular weakness, causing difficulty in swallowing food, which puts the child at risk of nutritional deficit. Continued use of this device requires careful attention and regular monitoring by nurses, focusing on infections, accidental aspiration and metabolic compromises[15].

The diagnosis of Aspiration Risk was listed using the nasoenteral tube[15], but mainly by the use of mechanical ventilator and tracheostomy. Muscle weakness characteristic of Pompe disease leads to respiratory dysfunctions, especially to diaphragmatic weakness[16]. Such conditions place the child in a condition of Risk for Impaired Breathing.

The risk of infection was indicated by the use of all invasive devices, and it is necessary for its prevention measures of adequate hygiene of the hands before carrying out any care with the child. It is recommended that the manipulation of the tracheostomy cannula, aspiration or dressing change be performed with modified clean technique[17].

Although the child presents priorities for care, it is essential to list, in this case, the diagnosis Risk of Child Development Impaired, considering the occurrence of retardation of psychomotor development. To meet this need, the health team, in addition to sending the child to specialized services, should periodically carry out home visits to teach and support the family to perform affective, physical, cognitive and sensorial stimuli, which will potentiate the development of the child[18].

Although the mother has taken care of the child through the guidance of hospital professionals, it is also necessary to offer her a systematic and specific care given the complexity of the situation experienced. The nurse can offer technical support to perform the essential care, in order to avoid complications, as well as in the adaptation of the child and family to the special needs.

The need to implement this daily care has a strong impact on the family structure, as it causes a burden on the caregiver and

financial difficulties. This family need eventually generated a demand for family care, which was expressed by six nursing diagnoses. Identifying these demands is essential to provide social support to the family, because if there is no social network that supports it, imbalances may occur, which will negatively influence the health of all members, especially the child and the family caregiver[14].

From the above, the health team should feel like a caring agent for these children and their families, allowing them a network of support and integration with other levels of health care. To this end, it is necessary to help the family members in their experiences, since hospital discharge, through interdisciplinary care networks, acting as facilitators of family empowerment for care[19].

The present case study and the elaboration of an intervention plan may provide support to professionals, especially nurses, to develop their skills and promote support and assistance to families. It is important to emphasize that it is not enough to determine the diagnoses and the interventions; it is necessary to apply them and evaluate their results for the continuity or change of actions, considering the objectives reached by the professional assistance. Therefore, it is considered relevant the permanent education to instrumentalize and qualify the teams in the care of people with rare diseases, guaranteeing integral and humanized assistance[6].

Moreover, in research with families, the literature has highlighted the use of the genogram and the ecomap, since these instruments allow us to know how families organize themselves for home care, the resources and support networks that they have to support them in the experience of chronic illness[20]. The use of these instruments allowed us to know the family composition and its network of social and professional support, essential factors for the planning of home care.

CONCLUSION

In carrying out this case study, it can be observed that children with Pompe disease have numerous needs, involving medication care, with technological, habitual and modified developmental devices. However, such care should expand to family needs, making them potentially ready to perform home care.

With respect to nursing care, the results showed that the classification of the practices allowed creating more specialized possibilities to elaborate and execute the care plan, based on the real needs of the children with Pompe disease and their relatives.

It is important to highlight the need for multiprofessional follow-up at the most important points of attention, since Pompe disease may impact the quality of life of patients and their families. Thus, the involvement and active participation of PHC professionals is essential to enhance the care of the children and their family.

Regarding the limitation of the study, the fact that a rare disease was addressed made it impossible to compare cases and generalize the results; however, the care plan developed from CIPE could be implemented to other people with a diagnosis of Pompe disease, safeguarding the particularities of each case.

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