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Nursing team in care of children with Pompe disease in intensive care

Equipe de enfermagem no cuidado à criança com doença de Pompe em terapia intensiva

Equipo de enfermería en la atención de niños con enfermedad de Pompe en cuidados intensivos

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ABSTRACT

Objective: to investigate nursing team knowledge and practices regarding care for children with Pompe disease in intensive care. **Method:** in this qualitative, descriptive study, data were collected by semi-structured interviews of nurses and nursing technicians working in the neonatal intensive care unit of a hospital in Rio Grande do Sul, after ethics committee approval. Data were subjected to content analysis. **Result:** the nurses emphasized experiences that go beyond technical procedures, in the endeavor to provide safe and qualified comprehensive care in order to provide experiences closer to a home for the children and their families. **Conclusion:** the nursing team was knowledgeable about care and worked in multidisciplinary manner. It was concluded that studies relating to rare disease offer input to inform nursing care.

Descriptors: Glycogen Storage Disease Type II; Metabolism, Inborn Errors; Intensive Care Units, Neonatal; Nursing Care.

RESUMO

Objetivo: investigar o conhecimento e práticas da equipe de enfermagem em relação ao cuidado à criança com Doença de Pompe em terapia intensiva. **Método:** trata-se de um estudo descritivo com abordagem qualitativa. A coleta de dados foi realizada com entrevista semiestruturada com enfermeiras e técnicas em enfermagem que atuavam na Unidade de Terapia Intensiva Neonatal de um hospital do Rio Grande do Sul, após aprovação pelo Comitê de Ética em Pesquisa. Os dados foram analisados pela análise de conteúdo. **Resultados:** as profissionais enfatizaram experiências que superam procedimentos técnicos, na busca de fornecer um cuidado integral qualificado e seguro, para proporcionar vivências mais próximas de um lar para a criança e familiares. **Conclusão:** a equipe de enfermagem possui conhecimentos para o cuidado e atua de forma multiprofissional. Conclui-se que estudos relacionados às doenças raras oferecem subsídios para qualificar o cuidado de enfermagem.

Descritores: Doença de Depósito de Glicogênio Tipo II; Erros Inatos do Metabolismo; Unidades de terapia intensiva neonatal; Cuidados de enfermagem.

RESUMEN

Objetivo: investigar el conocimiento y las prácticas del equipo de enfermería sobre el cuidado de niños con enfermedad de Pompe en cuidados intensivos. **Método:** en este estudio cualitativo descriptivo, los datos fueron recolectados mediante entrevistas semiestructuradas a enfermeras y técnicos de enfermería que laboran en la unidad de cuidados intensivos neonatales de un hospital de Rio Grande do Sul, previa aprobación del comité de ética. Los datos se sometieron a análisis de contenido. **Resultado:** las enfermeras destacaron experiencias que van más allá de los procedimientos técnicos, en el afán de brindar una atención integral segura y calificada con el fin de brindar experiencias más cercanas a un hogar para los niños y sus familias. **Conclusión:** el equipo de enfermería tenía conocimiento del cuidado y trabajaba de manera multidisciplinar. Se concluyó que los estudios relacionados con las enfermedades raras ofrecen información para informar la atención de enfermería.

Descriptores: Enfermedad del Almacenamiento de Glucógeno Tipo II; Errores Innatos del Metabolismo; Unidades de Cuidado Intensivo Neonatal; Atención de Enfermeira.

INTRODUCTION

Pompe Disease (PD) was discovered in 1932 by pathologist Joannes Cassianus Pompe, during the autopsy of a seven-month-old child who died from idiopathic myocardial hypertrophy. At the opportunity, the pathologist visualized glycogen accumulation in vesicles inside the cardiac fibers[1]. The pathology, also known as type II glycogen storage disease or acid maltase deficiency, is characterized by being a recessive, autosomal, progressive and multisystemic neuromuscular disease, caused by the accumulation of intralisosomal glycogen, by the ineffective activity of the Acid Alpha-Glycosidase (AAG) enzyme[2].

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Three types of PD are recognized in the literature, which are differentiated by severity and age group of manifestation, with classical childhood onset, non-classical onset and late onset. In classical childhood onset, the signs and symptoms appear in the first months of life, evidenced by myopathy, hypotonia, inappropriate growth, dysphagia, dyspnea and hearing disorders[3]. In Europe, the prevalence of the disease is approximately one case per 283,000 inhabitants and it is also known that, when manifested in childhood, only 25% of the children remain alive until one year of age[4]. In Brazil, the first case of the child form of PD was treated and published in 2008[5].

Advances in relation to early diagnosis are recent, such as the possibility of identification by the heel prick test, with biochemical investigation of autosomal genetic diseases, such as PD, Gaucher disease and Fabry disease. As soon as the diagnosis is defined, it is fundamental to start treatment with a team of professionals experienced in the care of PD. An essential aspect is genetic counseling as a component of care and a source of subsidies about the disease for patients and family members, which will help in current and future decisions[6].

Currently, the treatment of the disease is carried out with the implementation of continuous enzyme replacement therapy, combined with palliative support, inclusion of physical activities, physiotherapy, diet care, adequate pharmacotherapy and ventilatory support in cases of greater cardiac or pulmonary impairment. Thus, it is possible to provide greater quality of life and functionality for those affected by the pathology. The progression of the disease inevitably develops over the years[7]. With the enzyme replacement therapy, the estimated mean survival is 54% to 72%[8,9]. Alpha-glycosidase prolongs survival and quality of life in patients who may or may not need mechanical ventilation[10]. No matter how much the medication acts in the prolongation of the life estimate and regression of the clinical aggravation of the disease, it progresses towards the decline of the ability to perform daily activities and to lower quality of life[2].

In the Nursing practice, it is fundamental that the care provided to the child understands assistance and humanization to the patients and their family members during hospitalization, with the operationalization of welcoming and dialogical care[11,12]. In this perspective, the performance of the Nursing team is pointed out as a possibility to promote the improvement of the ability to meet the needs and care for children with PD and their family[13]. In this context, the role of the nurse is fundamental for her performance at the front line of care and with training based on holistic performance, in order to provide effective, safe, ethical and dynamic care planning[11].

Given the above, the following questions emerge: What is the knowledge of the Nursing team about PD? What is the role of the Nursing team in relation to children with PD in intensive care? Thus, the objective was to investigate the knowledge and practices of the Nursing team in relation to the care of children with Pompe Disease in intensive care.

METHOD

This is a descriptive research study with a qualitative approach, developed in a neonatal Intensive Care Unit (ICU) of a medium-sized hospital located in the Northeast of the state of Rio Grande do Sul[14]. Since 2011, the institution has been providing care to a child with PD, who is an inpatient of the institution until the present day, which justifies the intentional choice of the place for research.

The first contact with the institution occurred with the Responsible Technical Nurse, in order to present the research project and request authorization to carry it out. Upon approval to develop the research, it was presented at a sectorial meeting for the neonatal ICU Nursing team, composed of 19 nursing technicians and five nurses. At the end of the explanation, a list was provided to fill in the data of the team members interested in participating. Interviews with the team members were scheduled according to their availability.

The study participants were nurses and nursing technicians working in the aforementioned sector. The following inclusion criteria were used: being a member of the Nursing team working in the neonatal ICU, in any shift, and having provided care to a patient with PD. Exclusion criteria: being a nurse or nursing technician on leave of any kind or vacation during the data collection period.

After signing the Free and Informed Consent Term, in two copies, the semi-structured interviews were conducted on three days in the month of October 2019, in the morning, afternoon and night shifts, in a unique and individual manner, and in the workplace, respecting the participants' availability. This was followed by a script for collecting sociodemographic and work data with information on gender, age, work shift, time of professional practice and length of service in the institution. The specific questions about PD and care were based on the Genetics Home Reference, Your Guide to Understanding Genetic Condition[3].

The interviews were recorded using a digital recorder with authorization from the participants and transcribed in full for analysis, according to the method of thematic content analysis in the stages of pre-analysis, exploration of the

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material, and treatment of the results[15]. After exhaustive reading, data was organized with the coding of the results, the inference, extracting categories to, at a third moment, perform the interpretation and discussion of the data with the specialized literature on the subject matter. After carrying out the floating reading, the data were coded in registration units, which were systematically aggregated in two thematic categories: Knowledge of the Nursing team about Pompe Disease and its treatment; and Practices of the Nursing team in the care for children with Pompe Disease. In the data interpretation phase, the theoretical framework was resumed, in order to support the analyses giving meaning to the interpretation.

The ethical and scientific requirements for research studies involving human beings were ensured, through Resolution 466/2012 of the National Health Council, which sets forth the guidelines and regulatory standards for research involving human beings. The project was approved by the Research Ethics Committee, under CAAE No. 15690619.4.0000.5354 and opinion No. 3,428.965; and all the participating nurses were informed about the project and signed the Free and Informed Consent Form. To present the results, the participants were coded as Participant 1 (P1), Participant 2 (P2), and so on.

RESULTS AND DISCUSSION

The study participants were 11 professionals from the Nursing team, nine of them being nursing technicians and two nurses. Most of the participants were between 26 and 35 years old (63.63%), all were female, white race (100%), and most of them with a minimum time of performance of eight years in the Nursing area (72.72%). Two categories emerged from analyzing the speeches: Knowledge of the Nursing team on Pompe Disease and its treatment; and Practices of the Nursing team in the care for children with Pompe Disease.

Knowledge of the Nursing team on Pompe Disease and its treatment

It is known that PD is a rare pathology, which makes it difficult to access and spread knowledge on the theme. However, the team demonstrated to be aware of the importance of understanding about the pathology. Of the knowledge about PD approached by the Nursing team, that about the rarity of its occurrence, severity, chronicity, high lethality and systemic progression of the pathology was verified.

> *[...] It's a degenerative disease, in the muscles [...] it gradually left it flaccid, flaccid, so much so that when she came with us, she was five months old (patient), she grew up, she was still able to say goodbye with her hand. Now she doesn't do any of that anymore, she can only move her tongue and eyes, the rest is past. (P1)*
>
> *[...] has an enzyme deficiency [...] causes loss of all muscle strength, progressively. (P6)*
>
> *[...] when she was a baby with the passing of days she was unable to evacuate and breastfeed and they went to investigate, so they found out it was this disease. (P3)*

PD is an uncommon and gradual disease with high lethality. With the occurrence of the disease, the deficiency of the AAG enzyme emerges, as well as the need to replace it in a synthetic manner, in an attempt to mitigate the effects of the pathology progression and provide quality of life[16]. This treatment is continuous and is called Enzyme Replacement Therapy (ERT).

> *[...] she (child) lacks an enzyme, so she replaces it every 15 days. Because of this enzyme, she's alive. (P5)*
>
> *[...] doing the enzymes makes her more willing, happier. When she's getting close to doing the enzymes she starts to get more tired. (P3)*
>
> *The enzyme replacement therapy replenishes the enzyme, because Pompe occurs due to enzyme deficiency. With the therapy, we replace the enzymes and make the body break down the glycoside that is deposited in the organism. Treatment is with Myozyme®. (P9)*
>
> *[...] with gain weight, the number of bottles increases. (P11)*

Diseases like this, with no possibility of cure, are eligible for the modality of treatment with palliative care[17]. Palliative care aims to improve the quality of life of patients and family members through biopsychosocial assistance and, as with any other treatment, organization, planning and training are necessary. For the treatment of PD, ERT is performed with the administration of Human Recombinant Acid Maltase (alpha-glycosidase), with the trade name of *Myozyme®*[18]. Alpha-glucosidase is an effective treatment for the vast majority of patients with PD; however, this treatment offers no possibility of cure, which requires a thorough investigation of the individual response to the therapy[19]. ERT is a therapy that involves a large part of the multidisciplinary team, such as physician, pharmacist, nurse and nursing technician. However, its preparation is carried out by the nurses, with an institutional protocol, which guides it.

> *[...] the nurse is monitoring. It's done intravenously, so it has a specific preparation. (P2)*
>
> *[...] it's done with an infusion pump, starts with a very low volume and gradually increases over time, then measures the temperature, increases the volume if everything is right. Monitoring is by checking the vital signs.*

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Her doctor is the one who prescribes, and keeps follow-up, the pharmacy separates the day before and leaves it for the nurses to dilute. (P6)

A case study carried out in 2016 reported that the scientific community maintains consensus on the effectiveness of ERT, in addition to providing evidence of its benefits for the patient's clinic condition, positively contributing to the stabilization and motor development of the patient in the case of child PD[20]. In such a way, it guarantees quality of life to the patients, while new studies are not developed in search of better treatments. Thus, it involves complex situations, the role of a multidisciplinary team being essential. The preparation of the ERT is essential and, to ensure the proper processing of the medication, it is necessary to follow a series of precautions during the dilution of the enzyme, so as not to compromise its stability and effectiveness, as mentioned in the statements below.

[...] to dilute it is left at room temperature, then for dilution it has to be very careful. It is done slowly with a 25 x 7 needle, distilled water, placed on the wall of the flask, then it is diluted with circular movements, so as not to make bubbles, aspirate from inside the flask very slowly, so that it does not break the enzyme molecules, and then put it in the saline bottle to make the infusion. It is very time consuming, very careful to be able to dilute and administer it. The equipment must be PVC-free and have a filter. (P9)

[...] we wash (the portocath central venous access) with 20 ml, make a flushing and leave it salinized for 24 hours. (P7)

The Brazilian Ministry of Health released a report of recommendations for administering ERT in PD. Among its guidelines, the indicated dosage of alpha-glucosidase is 20 mg/kg, which must be infused every 14 days, through an intravenous route. The determination of the total volume varies according to body weight, with a mean infusion time of four hours in an infusion pump. Also, a prefilter (0.2 µ) must bate, respecting the limit of the infusion rates. The maximum initial infusion rate is 1 g/kg/h, which can reach 2 mg/kg/h every 30 minutes, after ensuring that the patient is stable, tolerating the medication, and then increasing the flow rate until the maximum of 7 mg/kg/h is reached. The Nursing team checks the vital signs at the end of each stage, always before proceeding to the next infusion rate[18].

The adverse effects reported by the team are not specific only to this ERT, but are general allergic reactions. The Nursing team demonstrates the necessary knowledge to act in situations of adverse effects of the medication, in addition to maintaining constant monitoring and evaluation of the patient, with previously defined strategies.

[...] we are monitoring to see if there is no fever, monitoring her. (P2)

[...] can make an allergic reaction, anaphylactic shock. (P11)

[...] some redness, blisters on the skin, skin reaction. (P3)

[...] in case of reaction we inform the nurse and she communicates the doctor on duty. Always have epinephrine with a prescribed dose. (P6)

Among the adverse reactions that can occur, anaphylactic reactions during the infusion stand out. However, the reactions that most occur and that require interventions are bradypnea, tachycardia, tachypnea, cough, vomiting, itching, skin rashes and fever[18]. The Nursing team reported the rarity, chronicity, severity and progression of the pathology, in addition to identifying the characteristic signs and symptoms of PD, such as hypotonia and motor impairment. They also demonstrated necessary knowledge on ERT, its benefits, possible adverse effects and necessary interventions in these cases. This indicates the involvement of the Nursing team in seeking theoretical support to improve care for patients with PD.

Practices of the Nursing team in the care for children with Pompe Disease

In their statements, the members of the Nursing team showed themselves committed, affectively involved, with emotion, affection, love and pride for having the opportunity to care for a child with PD. The professionals emphasized experiences that go beyond technical procedures, in the quest to provide qualified and safe comprehensive care, so as to provide experiences closer to home for the child and the family members.

[...] we have to take care to lateralize, the arms have to be taken care of, not to be left folded, the legs have the pillows that you put below! And even the socks we have to be careful not to leave them folded! Even the oximeter, the wire, everything carefully. (P5)

[...] she has comprehensive care that, as I see it, is wonderful, we have a nursing technician who stays only with her. (P10)

[...] she requires a lot of care! All her movements receded. I've taken care of her since she came here, baby. She moved, the little hands, the little legs, she would say goodbye, even a few words would come out, with time this went backwards gradually. We created a bond, she's like family, she's lives with us, right! She sees and is already flirtatious, interacts with the look. (P2)

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It is known that Nursing is the science and art that assists human beings in their basic human needs, with a view to making them independent by means of education to recover, maintain and promote their health[21]. Nursing care for patients with PD must meet all their needs, as highlighted in the statements that follow.

> *[...] aspiration and hygiene care, tooth brushing, bed bath, diaper, decubitus change every two hours or even less! She doesn't move, so, whatever position you leave her, she stays, you have to be careful! The diet too: washing the jejunostomy tube, medication via tube, hydration, aspiration, I don't know if I talked about the tracheo? The airways! (P3)*

> *[...] her respiratory rate, she uses a respirator! She uses the portable respirator to stroll here in the hospital, in the wheelchair, all tidy. That helps her! (P5)*

Mechanical ventilation is linked to high rates of pneumonia, so measures have been developed to prevent Mechanical Ventilation Associated Pneumonia as actions that address such demand. This set includes measures such as: keeping the headboard elevated from 30 to 45 degrees; aspirating the subglottic secretion routinely; and performing oral hygiene with antiseptics[22].

Another important factor in relation to Nursing care is the way in which communication is carried out between the child and the Nursing team. Strategies were created to enable communication with the patient in the progressive process of the pathology, since verbal communication is not possible, as well as non-verbal communication is compromised by immobility and generalized muscle atrophy.

> *[...] she manages to express what she needs with her face, and as we have been with her for a long time, we manage to understand what she wants! (P10)*

> *[...] she interacts, she responds to the stimulus, you talk to her. You say: do you want to stay there? Do you want to turn this way? We ask if she wants the lipstick, if she does she makes the little sign. She has a little sign like this, moving her mouth once means yes, moving backwards is no. So, she manages to interact in her own way, very restricted but we manage to have some communication. Then her teacher comes and talks, about some things and asks: do you understand? She answers with her little mouth, whether yes or no. We manage to understand her. (P3)*

Attention to the subjects in a holistic manner, which contemplates them in their biopsychosocial constitution, needs to ensure effective communication. A research study developed with the objective of analyzing the concept of comfort in a pediatric ICU, from the perspective of the Nursing professionals, led to the reflection regarding the Nursing care that is provided to hospitalized children. Addressing comfort in the face of all the technology and communicating with children brings benefits to everyone involved in this scenario[23]. This statement reinforces the commitment of Nursing to the needs of human beings with quality of life.

From the perspective of comprehensive care, the Nursing process guides and qualifies care, allowing the Nursing team to constantly assess the patient's health status, identify problems, plan interventions and evaluate the conducts performed, in addition to ensuring the record of the care provided, as can be perceived in the statements.

> *[...] of the nursing diagnoses she has: risk of skin integrity; risk of urinary tract infection; risk of bleeding due to aspiration of the tracheostomy; risk of infection; risk of insufficient nutrition. (P5)*

> *[...] in the system there is a list that we created, it includes the risks and diagnoses with interventions relevant to the patient. (P9)*

In child care, Nursing Care Systematization (NCS) provides effective reception of the child and family members during hospitalization, due to the understanding that nurses have in their praxis and scientific knowledge[24]. It has been proved that NCS promotes permanent and continuous improvements in the institutions, even when related to institutional management, in addition to qualifying assistance and management of work processes[24]. The care reported by the Nursing team is focused on promoting humanized care, considering the biopsychosocial aspects, with the multidisciplinary team acting together with family members, in order to promote quality of life in children with PD.

Study limitations

The study had some limitations, such as having been developed in a single private institution, with a single Nursing team, although this team has been assisting a PD patient for approximately eight years, which made it possible to develop strategies to qualify assistance, in addition to monitoring the progression of the pathology. This limitation was due to the rarity of the pathology and the opportunity for intervention with treatment. Continuous and in-depth studies are needed due to the complexity of the topic and its impact on society, with an emphasis on patients, family members and professionals involved in the process of PD progression.

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FINAL CONSIDERATIONS

The study demonstrated that the Nursing team has the necessary knowledge about the pathology, integrating with the other professions to provide comprehensive care to children with PD. The care performed by the team goes beyond Nursing care practices and extrapolates the basic care developed daily in a neonatal ICU, for not considering only the pathology, but also the basic human needs. In such a way, what the team demonstrated through the speeches, more than techniques and routines, seen as the basis of Nursing, were statements loaded with empathy and, above all, equality, adequate for the development phase of the patient being cared for.

Publications on PD for the Nursing area are still scarce. Research studies on the topic are suggested, which can assist both technical performance and Nursing care management. The publication of studies on Nursing care for patients with rare diseases has the potential to promote health in the patient and favor excellence in care.

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