

Practice of care in nursing in the McCune-Allbright Syndrome

Práxis do cuidar em enfermagem na Síndrome McCune-Allbright

Práctica de cuidado en enfermería en el Síndrome de McCune-Allbright

RESUMO

Objetivo: descrever os principais diagnósticos e intervenções de enfermagem de uma pessoa idosa com a síndrome McCune-Allbright. Método: pesquisa qualitativa do tipo estudo de caso único, realizada a partir da análise do prontuário, no período de 01 de agosto a 10 de dezembro de 2022. Resultado: A acromegalia ocasionada pelo distúrbio da tireóide e disfunção hormonal levaram aos diagnósticos de distúrbio na imagem corporal, síndrome da dor crônica, risco de síndrome do desequilíbrio metabólico, risco de quedas e síndrome do idoso frágil. As intervenções foram fundamentadas na Classificação das Intervenções de Enfermagem e orientadas a partir do autocuidado e avaliação clínica. Conclusão: considera-se que o cuidado a doenças raras é complexo pela dificuldade de achados literários, corroborado pelo envelhecimento da pessoa idosa, sendo necessário contribuir na construção de conhecimento técnico-científico para a prática do enfermeiro aos usuários com demandas endócrinas.

DESCRIPTORIOS: Doenças do Sistema Endócrino; Doenças da Glândula Tireóide; Idoso; Diagnóstico de Enfermagem; Cuidados de Enfermagem.

ABSTRACT

Objective: to describe the main diagnoses and nursing interventions of an elderly person with McCune-Allbright syndrome. Method: qualitative research of the single case study type, carried out based on the analysis of the medical records, from August 1st to December 10th, 2022. Result: The acromegaly caused by the thyroid disorder and hormonal dysfunction led to the diagnosis of a disorder in the body image, chronic pain syndrome, risk of metabolic imbalance syndrome, risk of falls and frail elderly syndrome. Interventions were based on the Classification of Nursing Interventions and guided from self-care and clinical evaluation. Conclusion: it is considered that care for rare diseases is complex due to the difficulty of literary findings, corroborated by the aging of the elderly, and it is necessary to contribute to the construction of technical-scientific knowledge for the practice of nurses with users with endocrine demands.

DESCRIPTORS: Endocrine System Diseases; Diseases of The Thyroid Gland; Elderly; Nursing Diagnosis; Nursing Care.

RESUMEN

Objetivo: describir los principales diagnósticos e intervenciones de enfermería de un anciano con síndrome de McCune-Allbright. Método: investigación cualitativa del tipo estudio de caso único, realizada a partir del análisis de las historias clínicas, del 1 de agosto al 10 de diciembre de 2022. Resultado: Acromegalia provocada por trastorno tiroideo y disfunción hormonal llevó al diagnóstico de trastorno en el organismo imagen, síndrome de dolor crónico, riesgo de síndrome de desequilibrio metabólico, riesgo de caídas y síndrome del anciano frágil. Las intervenciones se basaron en la Clasificación de Intervenciones de Enfermería y se orientaron desde el autocuidado y la evaluación clínica. Conclusión: se considera que el cuidado de las enfermedades raras es complejo por la dificultad de los hallazgos literarios, corroborado por el envejecimiento de los ancianos, y es necesario contribuir a la construcción del conocimiento técnico-científico para la práctica de los enfermeros con usuarios con demandas endocrinas.

DESCRIPTORIOS: Enfermedades del Sistema Endocrino; Enfermedades de la Glándula Tiroides; Anciano; Diagnóstico de Enfermería; Cuidado de Enfermera.

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Estudo de Caso EN

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INTRODUCTION

McCune-Albright Syndrome (MAS) is a rare genetic disease, originally diagnosed in 1936 by Donovan McCune and in 1937 by Fuller Albright by the triad: Bone fibrous dysplasia, café-au-lait skin spots and precocious puberty, whether or not associated with other endocrinopathies with the original triad. 1 MAS is caused by somatic activating mutations of the GNAS (Guanine nucleotide binding protein, alpha stimulating) gene resulting in cellular hyperplasia and hypersecretion of growth hormone and thyroid hormones.²

It is classified as a functioning pituitary adenoma, some of them resulting from prolactinomas³⁻⁴, activating mutation of a subunit of the GNAS gene that stimulates the growth and function of osteoblasts, melanocytes and endocrine glands. This condition influences the development of organs and tissues, and affects endocrine glands, bones and various tissues.⁴

They also include an increase in insulin-1 growth factor (IGF-1), which participates in the regulation of bone growth, working together with increased levels of growth hormone (GH), the latter being related to other diseases such as hyperthyroidism, estrogen-producing ovarian cysts, Cushing's syndrome and renal phosphate wasting with or without

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The most frequent detection of MAS occurs through the dermatological clinical picture, from macules coffee with milk, with irregular and unilateral borders⁵, peripheral precocious puberty, more evident in females.¹⁻³⁶

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rickets/osteomalacia and acromegaly.³⁻⁴

Rare disease, with a prevalence of 1/1,000,000 cases in the general population³, it can lead to difficulty in screening due to the unfamiliarity of health professionals with the disease, which can have long-term compromises, especially in the elderly, who undergo significant changes in their metabolism.⁷

Nursing has a fundamental role in monitoring and assisting at all stages of the life cycle, keeping a close eye on individual subjectivities, for the early diagnosis of MAS and limitation of damage and actions for health promotion and prevention of diseases and injuries, recovery and rehabilitation of health.⁷⁻⁸ In this sense, the work aims to describe the main nursing diagnoses and interventions of an elderly person with McCune-Allbright Syndrome.

METHODS

This is a case study, which is the study of the particularity and complexity of a single case⁹⁻¹⁰, carried out through the analysis of the medical records of a user with the syndrome in question from the qualitative and descriptive research, fragment of the umbrella project entitled "Nursing care for the elderly with endocrinological demands".

The study was carried out with 1 user with McCune-Allbright Syndrome.

me, in a state public institution in Rio de Janeiro, specialized in endocrinological care, with data collection carried out between August 1st and December 10th, 2022. The medical record was used as a data collection instrument, where information about the anamnesis, history of the current illness, past pathological history and family history of the user were selected.

After data collection, thematic analysis was performed ¹¹, being organized from the endocrine disorders found in the medical record of the user with MAS, along with the main diagnoses, Nursing interventions and expected results.

The anonymity of the participant was guaranteed, not exposing his identity, meeting all the requirements in research with humans and being approved by the Opinion of the Research Ethics Committee No. 5,814,509.

RESULTS

Female user, 61 years old, referred to a specialized unit in 1995, coming from an outpatient care unit in Freiburg, where she had been monitored for hyperthyroidism since she was 15 years old. In 1992, she presented a picture of growth of the cranial box in the temporal parietal regions on the left, anterior occipital fossa. After 1 year of diagnostic analysis and because it is a rare disease, the diagnosis of McCune-Albright Syndrome was reached, and she was followed up for outpatient treatment. A lucid user, she brings in her anamnesis a complaint of headache, nausea with general malaise and a nodule in the skull region, without compromising her daily activities. In the past pathological history, she mentioned sexual precocity, menarche at 12 years old. At age 15, she was diagnosed with hypothyroidism, with diffuse goiter, which culminated in partial thyroidectomy. At the time, due to nodules and pain in the cranial vault, a computed tomography (CT) was requested,

Table 1: Interventions and results found in the diagnosis of Body Image Disorder, 2022.

NURSING DIAGNOSIS: DISTURBANCE IN BODY IMAGE	
CLASS: BODY IMAGE	DOMAIN: SELF-PERCEPTION
NURSING INTERVENTIONS CLASSIFICATION (NIC)	
<ul style="list-style-type: none"> - Assistance in self-care: essential life activities, cognitive stimulation, orientation towards reality and improvement in socialization, energy control, support for the caregiver and improvement in coping; - Orient the patient to normal changes in the body associated with various stages of aging, as appropriate; - Help the patient to discuss the stressors that affect body image due to congenital conditions, injury, illness or surgery; - Identify a way to reduce the impact of any disfigurement through clothing, wigs or cosmetics, as appropriate; - Help the patient to identify actions that improve appearance; - Facilitate contact with people with similar changes in body image. 	
EXPECTED NURSING OUTCOMES	
<ul style="list-style-type: none"> - Improvement of the patient's self-esteem and self-care, acceptance, body change and decreasing the rejection rate in the rise of the disease. 	

Source: Herdman, Kamitsuru¹⁵; Bulechek, Butcher, Dochterman¹⁶Johnson, Mass, Moorhead¹⁷. Data adapted by the authors, 2022.

Chart 2: Interventions and results found in the nursing diagnosis of chronic pain, 2022.

NURSING DIAGNOSIS: CHRONIC PAIN SYNDROME	
CLASS: PHYSICAL COMFORT	DOMAIN: COMFORT
NURSING INTERVENTIONS CLASSIFICATION (NIC)	
<ul style="list-style-type: none"> - Perform a complete pain assessment, including location, characteristics, onset/duration, frequency, quality, intensity and severity, in addition to precipitating factors; - Teach the use of non-pharmacological techniques; - Guidance on pharmacological methods of pain relief; - Determine the impact of pain experience on quality of life. 	
EXPECTED NURSING OUTCOMES	
<ul style="list-style-type: none"> - Provide pain relief, explaining to the patient about pharmacological methods with care and caution, ensuring quality of life. 	

Source: Herdman, Kamitsuru¹⁵; Bulechek, Butcher, Dochterman¹⁶Johnson, Mass, Moorhead¹⁷. Data adapted by the authors, 2022.

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which showed lesions suggestive of craniofacial fibrous dysplasia, associated with a pituitary gland tumor measuring 1.8 x 1.5 cm with compression of the optic chiasm. Social history: denies alcoholism and smoking. Family history: presents with diagnoses of arterial hypertension. On physical examination there were light brown macules on the back and buttocks, and galactorrhea. In laboratory tests, growth hormone - GH above normal values. Drug therapy was started with the aim of reducing the tumor volume, but the insulin-1 - IGF-1 growth factor remained high, even with the increase in the dose, and other drug therapies were adopted without success to suppress IGF 1, however, there was a decrease in the tumor. In a new magnetic resonance imaging, after 4 years of pharmacological therapy, there was no visible lesion in the pituitary region, anatomically shaped optic chiasm, but IGF-1 levels continued to increase. The medication was then replaced again, leading to rapid normalization of IGF-1, clinical improvement, without causing side effects. It does not have associated comorbidities such as Diabetes and Hypertension.

DISCUSSION

MAS is configured in the user in question by the presence of café au lait spots, hyperfunctioning endocrinopathy, craniofacial fibrous dysplasia and acromegaly¹², being identified the last signs in the case studied.

Fibrous dysplasia (FD) is a benign condition identified by the replacement of bone tissue by fibrous tissue, characterized by symptoms reported by the user, such as headache and pain, craniofacial deformities and proptosis (eyeball protrusion).¹³ FD results in severe craniofacial deformity, leading to the diagnosis of Nursing, disturbance in body image (Chart 1), as well as the diagnosis of Chronic Pain Syndrome (Chart 2).¹⁴⁻¹⁵

Table 3: Interventions and results found in the nursing diagnosis of metabolic imbalance, 2022.

NURSING DIAGNOSIS: RISK FOR METABOLIC IMBALANCE SYNDROME	
CLASS: METABOLISM	DOMAIN: NUTRITION
NURSING INTERVENTIONS CLASSIFICATION (NIC)	
<ul style="list-style-type: none">- Teach the use of non-pharmacological techniques to control nausea;- Control the environmental factors capable of evoking nausea;- Reduce or eliminate personal factors that precipitate or increase nausea;- Teaching about the use of prescribed effective antiemetic drugs;- Determine the patient's expectations regarding body image;- Help the patient separate appearance from feelings of self-worth.	
EXPECTED NURSING OUTCOMES	
<ul style="list-style-type: none">- Attenuation of pharmacological treatment and instructing the patient to use non-drug therapeutic methods may contribute to reducing the risk of polypharmacy.	

Source: Herdman, Kamitsuru¹⁵; Bulenckek, Butcher, Dochterman¹⁶ Johnson, Mass, Moorhead¹⁷. Data adapted by the authors, 2022.

Table 4: Interventions and results found in the Nursing diagnosis Risk for falls, 2022.

NURSING DIAGNOSIS: RISK FOR FALLS	
CLASS: PHYSICAL INJURY	DOMAIN: SECURITY/PROTECTION
NURSING INTERVENTIONS CLASSIFICATION (NIC)	
<ul style="list-style-type: none">- Recommendation of safe behavior;- Participate in physical activity programs, always aiming at the user's condition with the help of a specialized professional;- Improvements in environmental safety;- Guide the user about possible change in vision, making it impaired with the pre-existing disease, recommended performing the Snellen test in the Nursing consultation;-Wear shoes with non-slip soles;- Instruct the user to never wear socks at night.	
EXPECTED NURSING OUTCOMES	
<ul style="list-style-type: none">- Establishment of goals for the user with home care, thus reducing possible risks, improving physical condition and developing more motor aptitude and self-confidence.	

Source: Herdman, Kamitsuru¹⁵; Bulenckek, Butcher, Dochterman¹⁶ Johnson, Mass, Moorhead¹⁷. Data adapted by the authors, 2022.

The acromegaly presented by the user, in turn, is characterized by the excessive production of growth hormone, which can lead to changes in facial characteristics and headaches, as well as morbidities such as Diabetes, Cardio-respiratory diseases and Neoplasms.¹⁸ The diagnosis of risk of disturbance in body image and the Chronic Pain Syndrome also caused by FD is reinforced

here (Charts 1 and 2).¹⁴⁻¹⁵

One of the drug therapies used was to contain the increase in the cranial cavity, due to craniofacial DF, causing the progressive loss of bone mass, making the bones weakened and predisposed to fractures and osteoporosis. Furthermore, osteoporosis is common in women over 45 years old, configuring a risk due to age and the bone changes caused.¹⁹

Due to aging and the possible impairment of the user's vision related to a pre-existing disease, there is a need to propose in interventions the performance of visual acuity tests using the Snellen sign scale, in order to assess the patient's degree of difficulty. Thinking about the impact of drug therapy on the health of the user and the possible impaired vision, the diagnosis of risk of falls was identified (Chart 4).¹⁵

The rare and difficult-to-diagnose clinical picture is associated with the aging of the user, a process that encompasses structural, chemical and functional neuro-biological transformations. Nursing, in view of the unfavorable clinical picture, combined with signs and symptoms that compromise the well-being and quality of life of the case studied and considering the imbalance that affects the health of the elderly person, must identify the possible weaknesses of the user. Disability is one of the problems that may be associated with the diagnosis of Frail Elderly Syndrome (Chart 5).^{15,20}

It is believed that this care should be subsidized by the theory of self-care, developed by Dorothea Elizabeth Orem, defined as actions that the individual practices for their own benefit, which contribute to the prevention or treatment of health problems.²¹ Bearing in mind that care is longitudinal and ambulatory, the individual must receive nursing support in order to perform care to maintain independence, as an elderly person with MAS with compromised health due to hormonal

problems and the signs presented by the disease.

Parallel to direct health care, educational actions are essential for the development of self-care²², being fundamental the development of welcoming and bond formation for the effective transmission of information; assistance in possible obstacles to self-care, even if the user is encouraged and guided to practice self-care.

CONCLUSION

MAS is a rare and complex pathology with regard to Nursing care, due to the difficult diagnosis and multiple causes and few scientific studies, mainly focusing on the role of nurses in this case, making it difficult to care for users with this condition.

Based on the listed Nursing care, a careful and holistic look at users and their demands according to the evolution and manifestations of the pathology is essential. Offering non-pharmacological measures of comfort to the user, whether related to pain, nausea or stressors, can contribute to improving the quality of life.

The creation and maintenance of the professional-user bond and with their family members contributes to a good prognosis, since communication is fundamental in the treatment process if it is adhered to and done correctly by the user. Nurses should guide self-care and mediate it using the support-education system as a care tool, according to Dorothea Orem's theory.

This case study contributes positively to the construction of technical-scientific knowledge for the practice of Nursing for users with endocrine disorders, since MAS arises from a dysfunction in the thyroid. However, in view of the rarity of the disease, it is necessary to go deeper into the subject in order to improve Nursing care for users with MAS.

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MIS leads to a multicausal condition that leads to a hormonal imbalance and fundamental drug treatment for the control of biochemical markers of disease activity, GH and IGF-I.¹⁸ In view of the persistent physiological and hormonal imbalance, the risk diagnosis of Metabolic Imbalance Syndrome was listed (Chart 3).

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