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MARASMIC MALNUTRITION ASSOCIATED WITH CONGENITAL ADRENAL HYPERPLASIA: CASE REPORT

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Abstract: Objective: To highlight the importance of early recognition of children at risk of severe malnutrition, the implementation of interventions to reduce the complications of this condition, and the need for preventive strategies. Case description: An 8-month-old male infant with a 2-day history of loss of appetite associated with irritability was taken to the tertiary service of this study with a lack of subcutaneous tissue, sunken eyes, loss of Bichat's fat pad, and atrophy of the gluteal muscles. Weight: 4,265 g and height: 62 cm. Initial supportive measures were performed and tests were collected to investigate secondary causes. After initial stabilization, correction of specific nutritional deficiencies and gradual introduction of food through an orogastric tube was initiated in conjunction with the hospital's nutrition team. Laboratory investigation confirmed the diagnosis of Congenital Adrenal Hyperplasia as a secondary etiology. The patient was discharged after 13 days of hospitalization due to clinical improvement, with the establishment of oral nutrition and resolution of the respiratory condition, weighing 4,800 g and measuring 62 cm. Discussion: Severe malnutrition is a serious public health problem, leading to infections and impaired development. It is crucial to adopt a multidisciplinary approach to treatment, involving doctors, nutritionists and other professionals. To address the increase in pediatric malnutrition, governments and institutions must collaborate on prevention, diagnosis and treatment measures, in addition to implementing public policies and strengthening the health system. Analyzing cases such as the one described helps to understand and improve strategies to combat malnutrition.

Keywords: childhood malnutrition, marasmic malnutrition and congenital adrenal hyperplasia

INTRODUCTION

The Childhood Protein-Energy Malnutrition (PEM) is a condition that affects children on a global scale, mainly under the age of five and living in underdeveloped and developing countries. According to the World Health Organization (W.H.O.), there are 47 million children in the world, under the age of 5, in a state of malnutrition, representing 45% of deaths in this age group.[1]. When compared to a eutrophic child, a malnourished patient is 9.4 times more likely to die [2].

Specifically, in Brazil, according to the latest Household Budget Survey (POF; 2017-2018), the country has 10.3 million citizens with severe food insecurity, which corresponds to 5% of the population. A report from this same institution predicted that around 130 million people could suffer chronic hunger as a result of the Covid-19 pandemic[3].

This health crisis has generated an economic crisis, with a worsening of income and an increase in food prices, which has led to an increase in food insecurity, which has deepened the emergency of risks related to hunger.[4,5]. According to the Oswaldo Cruz Foundation, Brazil recorded in 2021 the highest rate of hospitalizations of children under one year old associated with malnutrition (113 hospitalizations for every 100,000 live births)[6].

The PED occurs due to an imbalance between the need for nutrients and their intake, which results in a cumulative deficit of energy, proteins and/or micronutrients, which can negatively affect the physical and cognitive development of the malnourished child. It can be classified as primary and secondary, with primary being caused by inadequate nutrient intake and secondary being caused by another pathology that generates a nutritional imbalance.[7]

By knowing these facts, it is essential to understand the extent and magnitude of this problem, requiring government measures to ensure access to adequate and healthy food for the population, in addition to a comprehensive approach to investigate possible etiologies and reduce morbidity and mortality associated with DEP.

In this article, we report a case of marasmic malnutrition in a child living in a city in the interior of northwestern São Paulo, referred to a tertiary hospital in the region. Through this, we emphasize the importance of early recognition of children at risk of severe malnutrition, the implementation of interventions to reduce the complications of this condition, and the need for preventive strategies.

MATERIALS AND METHODS

The data for this report were collected through analysis of the patient's medical records filed during the period he was hospitalized in the pediatric unit of a tertiary hospital in the northwest of São Paulo.

The Terms of Commitment for Use of Data and Medical Records, as well as the Free and Informed Consent Form, were duly completed and signed by the child's legal guardian.

To discuss the report, a bibliographic review was conducted using the keywords childhood malnutrition, marasmic malnutrition and congenital adrenal hyperplasia. The following databases were consulted: Scielo, PubMed, Cochrane and LILACS.

REPORT OF CASE

An 8-month and 2-day-old male infant presented with a 2-day history of loss of appetite associated with irritability, in addition to prostration after a choking episode during a meal. In his hometown, he received initial medical care that revealed signs of malnutrition, hyporeactivity, and hypoglycemia. Initial stabilization measures were performed with hypertonic glucose, ventilatory support, and oropharyngeal aspiration of food contents. However, the patient continued to show signs of instability. Due to these circumstances, it was decided to refer the infant to the pediatric emergency care unit of the hospital service where this study was conducted.

Upon admission, the patient was in poor general condition, pale 2+/4+, dehydrated 3+/4+, acyanotic, anicteric, afebrile, hypoactive, hyporesponsive, tearful, and emaciated 3+/4+. On physical examination, the respiratory system showed bilateral vesicular murmur with diffuse rhonchi and stertoration, tachypnea, presence of furcula retraction and intercostal retraction. The cardiovascular system showed no alterations. The abdomen was hollow, without visceromegaly, bowel sounds were present and painless on palpation. The patient presented scarcity of subcutaneous cellular tissue, sunken eyes, with loss of Bichat's fat pad and atrophy of the muscles of the gluteal region (Figure 1). Weight: 4,265 g (below the W/A Z score -3) and height: 62 cm (H/A Z score <-3). The other systems showed no alterations.

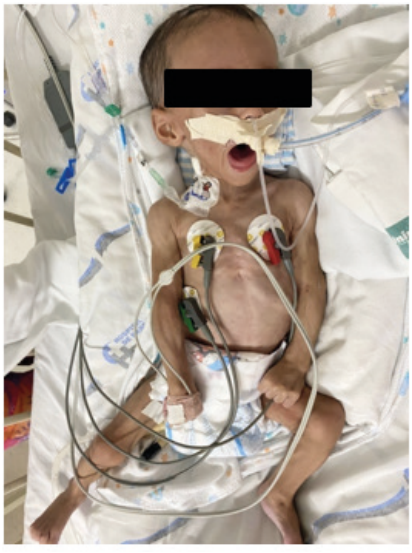

Figure 1: Clinical aspects of malnutrition at patient admission. Source: authors.

Laboratory tests on admission showed hypokalemia, positive initial infectious screening with leukocytosis and left shift in the blood count and increased C-reactive protein, prolonged activated thromboplastin time (APTT) and prothrombin time (PT), increased liver enzymes and arterial blood gas analysis with metabolic acidosis and hypoxemia. Initial blood and cerebrospinal fluid cultures showed no changes.

A chest X-ray was also requested for the initial evaluation, which showed bilateral hypotransparency with blurring of the lower cardiac contour, bilateral pulmonary hila, left apex and pinching of the costal arches in the left hemithorax.

Based on the patient's clinical presentation, a case of severe malnutrition was identified and the diagnosis of Marasmatic Protein-Energy Malnutrition was made, in addition to the association with a case of sepsis. Initially, due to the patient's serious condition, volume expansion and infusion of vasoactive drugs were performed to improve hemodynamics. Antibiotic therapy was also initiated to treat the sepsis, with a pulmonary focus associated with possible bronchoaspiration, mechanical ventilation support due to respiratory failure and decreased level of consciousness, basal maintenance serum, in addition to correction of the hydroelectrolytic disorders presented. After initial stabilization, correction of specific nutritional deficiencies and gradual introduction of food through an orogastric tube was initiated in conjunction with the hospital's nutrition team.

During the investigation of the primary etiology for the malnutrition, the companion reported that the patient was born prematurely and was being monitored in the city of origin by a pediatrician due to delayed neuropsychomotor development (NPMD). In addition, he did not have the patient's vaccination card and reported that he had undergone all neonatal screening tests after birth. Regarding the introduction of food, he reported that the patient received exclusively milk formula until 6 months, in quantities and preparation inadequate for his age, and from 6 months onwards he also began to receive complementary feeding with savory and fruit porridge, without offering all food groups, in addition to cow's milk with added sugar. She also reported that the infant was taking vitamins, but she was unable to give the name or dosage.

Secondary etiologies were also investigated during the follow-up. With the help of the hospital's social worker, who contacted the patient's original care service, it was discovered that the infant had not undergone the neonatal screening test and had an incomplete vaccination schedule for his age.

Furthermore, among the laboratory tests requested for the investigation, with congenital adrenal hyperplasia listed as the main hypothesis due to the clinical picture and social factor, the hormone precursor 17-alpha hydroxyprogesterone was requested, which presented an altered result of 1089ng/dL. Other tests, such as urinary sodium, thyroid hormones, and viral serological tests, were requested to rule out differential diagnoses such as metabolic and endocrine diseases, congenital infections, among others, all with normal results. Therefore, with the diagnosis of Congenital Adrenal Hyperplasia confirmed as the secondary cause of malnutrition, treatment was started with hydrocortisone 10mg/ml every eight hours and fludrocortisone 100mcg/day every twelve hours. The patient showed excellent nutritional recovery, as well as considerable improvement in mood and appearance, with the disappearance of the initial apathy (Figure 2). The patient was discharged after 13 days of hospitalization due to clinical improvement, with the establishment of oral nutrition

and resolution of the respiratory condition, weighing 4,800 g and measuring 62 cm (weight gain of 535 g = 41.1 g/day). The guardian who monitored the child's progress was instructed on an age-appropriate diet, regularization of the vaccination schedule, continued childcare follow-up at the Basic Unit of origin and, at the same time, began outpatient follow-up for specialized medical monitoring at the Hospital. Furthermore, the guardian was advised to seek medical attention if the child presented warning signs and/or seriousness.

Figure 2: Improvement in clinical aspects of malnutrition and weight gain during the patient's evolution. Source: authors.

DISCUSSION

Primary PED is the result of a complex interaction of economic and social events that lead to inadequate nutrient intake, while secondary PED is related to the presence of chronic or acute diseases that lead to compromised nutritional status due to various causes such as lung disease, heart disease, malabsorption syndromes, neuropathies, among others. [7,8].

It can present different stages, from the mildest forms to the most severe manifestations, such as Kwashiorkor (edematous malnutrition) and marasmus. Kwashiorkor generally occurs in patients between one and three years of age and has a higher fatality rate when compared to other clinical forms of malnutrition. [1]. In this type, there is a greater protein deficiency, leading to edema due to fluid retention generated by the rupture in weakened cell membranes, which causes ions to escape from the intracellular space to the extracellular space and, consequently, an increase in water flow.[8]. This way, the patient presents with pitting edema, mental and mood changes, generalized or localized changes in the scalp (flag sign), anorexia, diarrhea, infections and deficiencies (vitamin A, zinc and iron).)[9].

The marasmic form is more common in children under 12 months of age. In its clinical presentation, there is loss of muscle mass and subcutaneous fat; the patient usually presents with abdominal distension (due to muscle hypotonia), irritability, anorexia or signs of hunger, apathy, loss of Bichat's fat pad, favoring an aged appearance (senile facies), depressed anterior fontanelle, sunken eyes, loss of skin elasticity, chronic diarrhea and global delay in neuropsychomotor developmen[1].

In the case reported, we highlight that the patient presented marasmic malnutrition associated with nutritional deficiency and secondary to congenital adrenal hyperplasia.

Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency (D21OH) is a disease resulting from mutations in the CYP21A2 gene (or P450c21) responsible for the encoding and expression of the 21-hydroxylase enzyme in the adrenal cortex and essential for the biosynthesis of adrenal steroids. The absence or functional inactivity of this enzyme prevents the normal production of cortisol and aldosterone in up

to 75% of patients, diverting the accumulated intermediate products to the excessive synthesis of androgens.[10,11,12].

In the classic salt-wasting form, cortisol and mineralocorticoid production is compromised. As a result, the patient presents with decreased cardiac output, hydroelectrolytic disturbances, loss of appetite, lethargy, vomiting and inability to gain weight. [13,14].

Through neonatal screening, the diagnosis of CAH can be made early [15]. In addition, serum 17-hydroxyprogesterone (17-OHP) measurement is also a useful laboratory test for diagnosis. [16]. Treatment is based on glucocorticoid and mineralocorticoid hormone replacement. [17.]

In view of the above, it is essential that health professionals approach children comprehensively in order to reduce complications and mortality associated with this condition. The Manual for the care of children with severe malnutrition in hospitals, developed by the Ministry of Health, consists of three phases: stabilization of the condition, rehabilitation and monitoring.[19].

During the initial phase, problems that pose a risk of death, such as hypoglycemia, hypothermia, dehydration and infections, must be treated, specific nutritional deficiencies must be corrected, metabolic abnormalities must be reversed and feeding must be initiated gradually. [19, 20,21].

In the rehabilitation phase, which lasts approximately two to six weeks, intensive feeding is required in order to recover much of the lost weight, emotional and physical stimulation is provided, the caregiver is instructed to continue care at home and the child is prepared for discharge. In the third phase, the patient is referred for follow-up with the aim of prevention and continuation of treatment.[19,20].

CONCLUSION

Severe malnutrition continues to be a significant public health problem, widely associated with increased rates of infection, morbidity and mortality, and developmental impairment, as well as reduced quality of life. This case clearly illustrates the critical need for an intensive and comprehensive approach to the management of protein-energy malnutrition. The successful intervention demonstrated here underscores the importance of collaboration by a multidisciplinary team of professionals from diverse backgrounds, including doctors, nutritionists, nurses, psychologists, and social workers. Each of these professionals plays a vital role not only in nutritional rehabilitation but also in addressing the physical, emotional, and social needs of affected children.

With the increase in cases of malnutrition in the national pediatric setting, it is imperative that comprehensive measures be implemented in collaboration between different sectors of society. Governments, academia, social media, and other entities must join forces to promote prevention, early diagnosis, and effective treatment of malnutrition. The creation and implementation of robust public policies, such as the promotion of breastfeeding, education on healthy eating, and ongoing training of health professionals, are essential. In addition, strengthening the health system and ensuring that professionals are adequately trained to manage malnourished children are essential steps to address this challenge.

Reporting and analyzing cases such as the one described is crucial not only to raise awareness about malnutrition, but also to foster a deeper understanding of effective treatment and prevention strategies. Such efforts are essential to mitigate the socioeconomic and cultural repercussions of malnutrition and promote the health and well-being of affected children, contributing to a healthier and more sustainable future for society as a whole.

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