

Parenteral Nutrition Therapy for an Achondroplastic Patient: Case Report

Terapia nutricional parenteral para um paciente acondroplásico: Relato de caso

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Int J Nutrol 2019;12:61–65.

Abstract

Introduction Achondroplasia is the most common form of dwarfism in humans. At the end of the twentieth century, achondroplasia had its natural history investigated and its morbimortality understood. In dwarves, obesity is one of the causes of morbidity, and it is difficult to evaluate it due to the great disproportion among anthropometric data. The characterization of the nutritional needs of these patients represents an interesting dilemma for physicians. In view of these difficulties, the present case report describes an alternative to obtain the ideal weight value of an elderly dwarf in the use of parenteral nutritional therapy.

Case Report A 73-year-old male patient, carrier of achondroplastic dwarfism, was admitted for surgical treatment of colon adenocarcinoma. He was submitted to total colectomy with ileum-rectum anastomosis, evolving with abdominal septic shock by suture dehiscence, which was fixed, and a protective ileostomy was installed. Due to the maintenance of prolonged fasting, without the possibility of starting oral or enteral feeding, the patient used total parenteral nutrition. To estimate the total energy expenditure, we chose to ask the patient what his ideal weight was. The patient used total parenteral nutrition for eight days until he was able to meet his caloric needs orally.

Conclusion In the absence of indirect calorimetry, the ideal weight mentioned by the patient seems to be a good option for the estimation of the caloric expenditure by simplified equations.

Keywords

- achondroplasia
- energy expenditure
- ideal weight
- parenteral nutrition therapy

Resumo

Palavras-chave

- acondroplasia
- gasto energético
- peso ideal
- nutrição parenteral terapia

Introdução Acondroplasia é a forma mais comum de nanismo em humanos. No final do século XX, a acondroplasia teve sua história natural investigada e sua morbimortalidade entendida. Em anões, a obesidade é uma das causas de morbidade sendo difícil de ser avaliada devido à grande desproporção entre os dados antropométricos. A caracterização das necessidades nutricionais destes pacientes representa interessante dilema para médicos. Tendo em vista estas dificuldades, este relato de caso descreve uma alternativa para a obtenção do valor do peso ideal de um idoso portador de nanismo em uso de terapia nutricional parenteral.

Relato de Caso Paciente do sexo masculino, 73 anos, portador de nanismo acondroplásico admitido para tratamento cirúrgico de adenocarcinoma de cólon. Foi submetido a colectomia total com íleo-reto anastomose, evoluindo com choque séptico de foco abdominal por deiscência de sutura, a qual foi corrigida e confeccionada ileostomia protetora. Por manter jejum prolongado, sem possibilidade de início de alimentação por via oral ou enteral, paciente utilizou nutrição parenteral total. Para estimativa de cálculo do gasto energético total, optou-se por perguntar ao paciente o seu peso ideal. Paciente fez uso de nutrição parenteral total por 08 dias até atingir sua necessidade calórica por via oral.

Conclusão Na ausência de calorimetria indireta, o peso ideal referido parece ser uma boa opção para as equações simplificadas de estimativa de gasto calórico.

Introduction

Achondroplasia is the most common form of dwarfism in humans, and it was described hundreds of years ago; it currently affects around 250 thousand individuals worldwide.¹ It is a genetic condition characterized by shortening of the limbs, macrocephaly, middle facial hypoplasia, and small and wide hands.² It is believed to occur in 1/25 thousand live births.^{1,2}

Despite being a condition that has been long known, only at the end of the 20th century achondroplasia had its natural history investigated and its morbimortality understood. Hashmi et al³ described that the most common causes of death among achondroplastic patients have changed over the past 30 years, and are no longer pneumonia, hydrocephalus or sudden death. Today, most of these patients die from cardiovascular and cerebrovascular diseases or accidents.

Obesity is another frequent cause of morbidity in achondroplastic patients.^{1,4} However, it is difficult to evaluate this condition since the methods for the diagnosis of obesity do not enable the correct comparison between weight and height among dwarves, which is a great challenge.⁴ Due to the disparity found among the anthropometric data, the ideal weight estimation for these individuals seems to be an arduous task.⁵ In 1986, Hooks et al⁶ found that the characterization of the nutritional needs of a patient with achondroplasia represented an interesting dilemma for physicians.

Considering the difficulties of estimating the ideal weight for achondroplastic patients because of their short stature and the scarcity of studies on this issue, the following case report describes an alternative to obtain the ideal weight value and the consequent calculation of the caloric needs of an elderly patient with dwarfism using parenteral nutrition therapy.

Case Report

The present case report is about a male patient, aged 73 years, bearer of achondroplastic dwarfism and systemic arterial hypertension, weighing 53.6kg, with 107 cm in height, and body mass index (BMI) of 46.82 kg/m², who was admitted electively to the infirmary of our institution to undergo surgical treatment for a colon adenocarcinoma. He had been previously submitted to a prostatectomy to treat a

prostate cancer. Upon hospital admission, the patient was evaluated regarding his nutritional status using the Nutritional Risk Screening (NRS 2002) instrument, and was classified as free of risk.

On the first day of hospitalization, the patient was submitted to total colectomy with ileum-rectum mechanical anastomosis. The surgical procedure happened without abnormalities. After recovery from the anesthesia, he was sent to the Intensive Care Unit (ICU), hemodynamically stable, in spontaneous ventilation and awake. On the fifth postoperative day, the patient presented abdominal distension, fever, hypotension, tachycardia and leukocytosis, and was diagnosed with septic shock with an abdominal focus, and we decided to perform a new surgical approach. After an exploratory laparotomy, a punctual dehiscence of the anastomosis was identified, which was fixed, with subsequent performance of a protective ileostomy. The patient was once again referred to the ICU for postoperative evolution, this time intubated and in hemodynamic instability. A new evaluation of the nutritional risk was made using the NRS, which identified low risk of undernutrition for the patient. The patient underwent antibiotic therapy (piperacillin + tazobactam), with control of the blood pressure, and a gradual and progressive improvement of the condition was observed.

On the eighth day of hospitalization, the patient was not undergoing an oral or enteral diet, with improvement in the clinical hemodynamics, but he presented paralytic ileum, and it was not possible to start feeding. The Nutritional Therapy Service was asked to evaluate the case, and the introduction of parenteral nutrition therapy (PNT) was indicated. Due to the difficulty regarding the proper positioning of the central venous catheter in the jugular and subclavian veins, a right femoral vein puncture was performed. For the estimation of the total energy expenditure (TEE), since there is no calorimeter available in the hospital to conduct indirect calorimetry, and we did not find another standardized alternative for this procedure in the literature, we chose to ask the patient what he thought his ideal weight was (referred ideal weight – RIW = 40kg).

Thus, the TEE was calculated as 999 kcal (24, 9kcal/kg of RIW/day [d]), which were distributed as follows in the composition of the PNT: 60 g of amino acids (1.5 g/kg of

Table 1 Biochemical Exams

Exam	Beginning of the PNT	End of the PNT
Hb/Ht	8.6/25.7	8.8/27.1
AST	26	84
ALT	22	95
GGT	84	84
AP	39	75
TB/DB	0.40/0.20	0.40/0.20
Urea	108	57
Creatinine	3.8	0.7
Albumin	2.2	2.4

Abbreviations: ALT, alanine aminotransferase; AP, alkaline phosphatase; AST, aspartate aminotransferase; DB, direct bilirubin; GGT, gamma-glutamyltransferase; Hb, hemoglobin; ht, Hematocrit; PNT, parenteral nutrition therapy; TB, total bilirubin.

RIW/d), 135 g of glucose (3.37 g/kg of RIW and 2.34mg/kg of RIW/minute) and 30 g of lipids (0.75 g/kg of RIW/d) associated with electrolytes, vitamins and trace elements. The patient needed to undergo the PNT for eight days until reaching his TEE through oral intake, maintaining good glycemic levels and without alterations in the laboratory exams during this period, as shown in ►Table 1.

After the PNT and the withdrawal of the femoral catheter, the patient presented right lower limb edema. He was submitted to a colored venous Doppler of the limb, with identification of deep venous thrombosis. The patient underwent prophylactic anticoagulation for full anticoagulation.

On the 30th day of hospitalization, the patient was discharged; he was on a full oral diet, kept the use of the oral anticoagulant in alternated days, and a readmission for correction of the ileostomy was scheduled.

Discussion

Basal energy expenditure (BEE), the major component of the TEE of an individual, can be estimated with some accuracy through the equation proposed by Harris-Benedict. It takes into consideration the patient's age, height, weight and gender. In 1986, Hooks et al⁶ claimed that the usability of this formula for people with short stature had not yet been well established, which seems to be true even today. In 1990, Owen et al⁷ found that patients with achondroplasia had a higher basal caloric intake per weight unit than adults without the condition. In addition, they observed that BMI, skinfolds, or other anthropometric measures would be unable to predict the body composition of these individuals. They concluded that there is no way to estimate energy expenditure for dwarfs, and when nutritional therapy is needed, it would be best to measure the specific needs of each patient.

The methods for managing BEE are direct and indirect calorimetry. In our service, we do not have an indirect calorimeter available. Therefore, to obtain an adequate weight estimate, we opted to use the ideal weight referred by the patient himself. Comparing this information with the weight curve for specific age for children with achondroplasia (►Fig. 1) prepared by Hoover-Fong et al⁸ in 2007, we identified that 16-year-old dwarfs should weigh ~ 40 kg in the 5th percentile.

If we consider the BMI for the age curve in children up to 16 years old (►Fig. 2) made by Hoover-Fong et al⁹ in 2008 to compare the patient's BMI according to his RIW (34.94 kg/m²), we would observe that this would be above the 95th percentile. Therefore, the weight could be overestimated. This is probably due to the existence of a great variation of stature within the same age group. It is important to consider that there may be physiological anthropometric differences between 16-year-old achondroplastic teenagers, who are almost at the final stage of development, and elderly dwarves, such as the patient described in the present report.

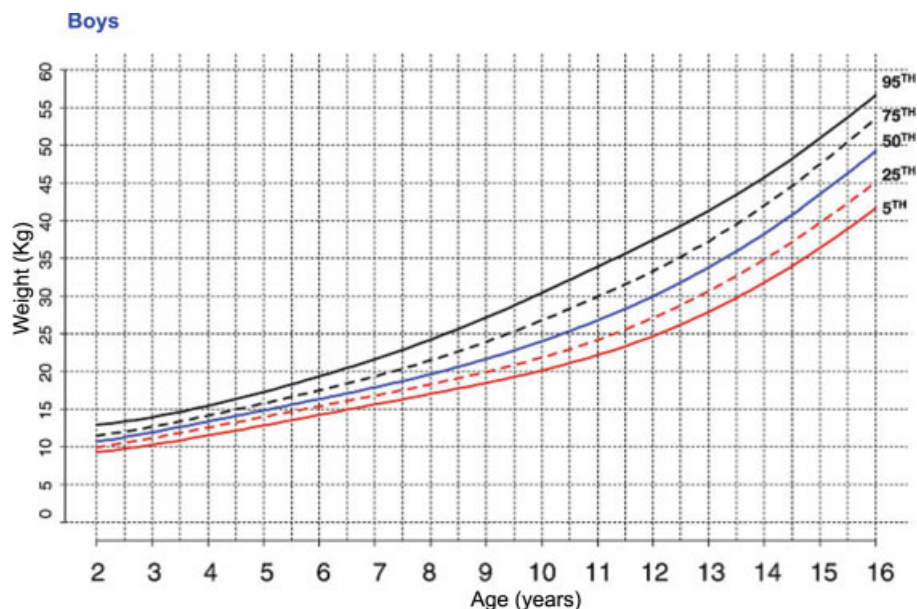


Fig. 1 Age-weight curve for achondroplastic children developed by Hoover-Fong et al.⁸

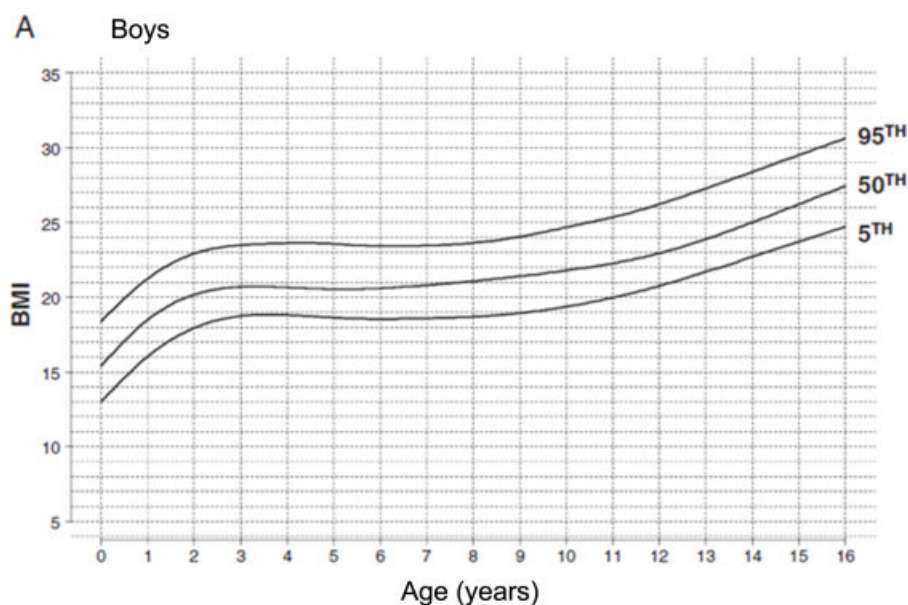


Fig. 2 Age-body mass index (BMI) curve for achondroplastic children developed by Hoover-Fong et al.⁹

When we compared the RIW of this patient with the curve developed by Del Pino et al¹⁰ for Argentine children and adolescents aged ≤ 18 years (**Fig. 3**), we found that dwarves in the final stage of development in the 50th percentile should weigh ~ 40 kg. This data was also compatible with the information suggested by the patient.

Checking the study by Merker et al,¹¹ the ideal weight reported by our patient would be below the average found for Caucasian achondroplastic adult men aged 20 years, which would be 54.5 kg. These differences show that there may be great variability among anthropometric data for dwarf individuals of different regions, and it is not possible to estimate an ideal weight for age, for example, through non-specific curves for a given population.

Regarding the calculation of calorie intake, McClave et al¹² stated that, in the absence of indirect calorimetry, it seems acceptable to use equations based on the weight (25–30 kcal/kg/d) to estimate the energy expenditure for critically-ill patients. With the use of the simplified formula in this specific case, we were able to estimate an appropriate energetic value based on the RIW. This is better evidenced by the fact that the results of the patient's biochemical exams did not present alterations suggestive of hypo or hyperalimentation.

Regarding the deep venous thrombosis developed by the patient, it is known that the femoral site itself increases the risk of this complication, regardless of the time the catheter stays there. Despite this, in cases in which there is any difficulty in the catheterization of the superior veins,

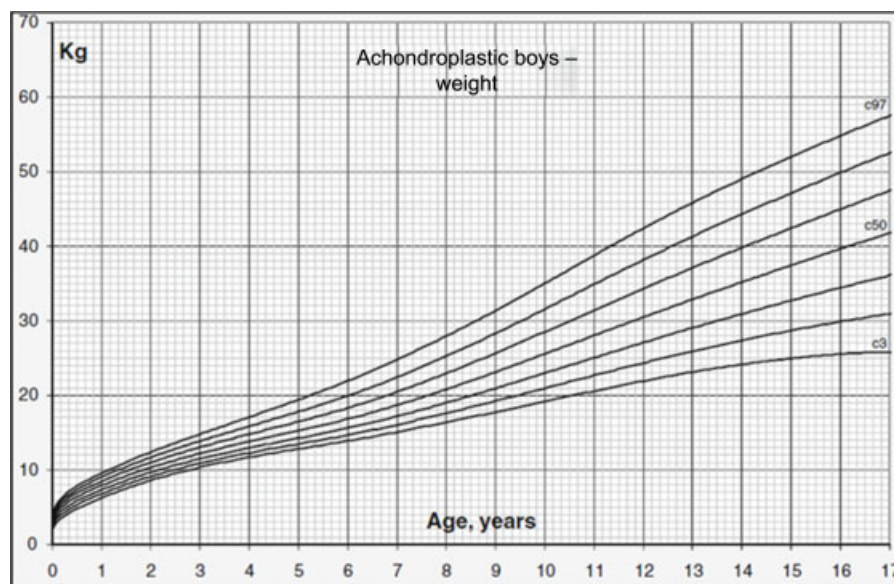


Fig. 3 Age-weight curve for achondroplastic children developed by Del Pino et al.¹⁰

especially in critical patients, the femoral site is still an important alternative, and it should be used.¹³

Conclusion

Achondroplastic dwarfs are challenging patients for physicians, especially regarding the mean weight for a given height and age. The methods used for individuals without this condition should not be applied to this specific population. In the absence of indirect calorimetry, the RIW seems to be a good option for the simplified calorie expenditure estimation equations. Further studies are still needed to determine the validity of this alternative.

Conflict of Interests

The authors have none to declare.

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