Importance of Early Diagnosis and Treatment in Cases of Phenylketonuria

Silva K.B¹, Pinheiro M.M.A², Filho S.L.V.N³

1Nutricionista Residente em Oncologia e Cuidados Paliativos ASCES UNITA; 2Nutricionista Graduada pelo UNIFAVIP/DeVry; 3Nutricionista Mestrando no Programa de Pós-Graduação em Saúde as Criança e do Adolescente do Centro de Ciências da Saúde-UFPE

**ABSTRACT**

Introduction: Phenylketonuria is a hereditary disease characterized by the absence of the enzyme phenylalanine hydroxylase, which converts the amino acid phenylalanine to tyrosine. Phenylalanine, accumulates in the body, and confers toxicity mainly to the central nervous system. Diagnosis, as well as early treatment is essential for the child to develop normally. Objective: To describe the diagnosis and treatment of phenylketonuric patients. Methodology: A review of the literature was conducted through articles published in journals indexed in the Scielo and PubMed databases, in the Portuguese language, in the last 8 years, using the descriptors: diagnosis, phenylketonuria, treatment. Results and Discussion: The diagnosis of phenylketonuria is made through neonatal screening, foot test, where the concentrations of phenylalanine and tyrosine are identified. When phenylalanine levels are elevated and tyrosine levels are low, the diagnosis of the disease is proven. When performed early, the diagnosis and consequently the start of treatment, can prevent the irreversible neurological damage that high doses of phenylalanine cause in patients. The recommended treatment is still a restricted diet of phenylalanine, with the use of a diet with low intake of phenylalanine in the child’s diet, in the first month of life, can significantly prevent mental retardation. That is, a feed free of foods containing phenylalanine protein, such as food of animal origin (meats and their derivatives) vegetable origin (legumes and oilseeds) and sweeteners and / or foods containing aspartame. The greatest difficulty in the elaboration of these diets is to meet the nutritional needs of the patient so that it has its full development. Conclusion: Early diagnosis along with adequate treatment will allow the patient with phenylketonuria to develop and grow optimally. The monitoring of the nutritionist and the physician, is of paramount importance to minimize the adverse effects resulting from the accumulation of phenylalanine.

**Keywords:** Diet; Nutrition; Phenylalanine; Phenylketonuria