Phenylketonuria (PKU; MIM 261600) is the most frequent inborn metabolic disease of amino acid metabolism. After diagnosis, actually made by the newborn screening, a natural protein and phenylalanine (Phe) restricted diet, simultaneously supplemented with a Phe-free amino acid mixture and special low protein foods is implemented. This nutritional approach has shown tremendous success preventing mental retardation. However, nutrition in PKU is a challenging issue since important imbalances may occur. While no consensus exists about the ideal protein intake, micronutrient status usually reveals extreme findings, underlining the need to carefully monitor nutritional status in these patients. The precise definition of the protein nutritional status in PKU is an old wish of clinical professionals. While the best biochemical marker is not yet defined, plasma prealbumin seems to be useful to decide the adequate protein intake in patients with PKU. Protein intakes according to the actual guidelines for patients with PKU seem to result in normal growth and body composition comparing to controls. Typical diet used to treat patients with PKU is restricted in natural protein but usually rich in carbohydrates. Despite these potential obesogenic diet characteristics, dietary treatment in PKU does not increase the risk of overweight, central obesity or increased body fat. In addition, patients with PKU reveal a tendency for reduced metabolic syndrome prevalence, although an atherogenic dyslipidemia was found. Although these patients with central obesity do not manifest an increased pro-inflammatory status compared to controls, future studies will be needed to help clarifying their long term cardiovascular risk, considering their atherogenic dyslipidemia whilst presenting reduced total cholesterol and glucose concentrations.

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