Abstract

Objectives: Assessment of the quality of dietary treatment of phenylketonuria (PKU) patients and investigation of its relationship with the general intelligence of the patients. Methods: Cross-sectional and longitudinal study of 105 PKU treated patients. The index of dietary control (IDC) was calculated as the phenylalanine (Phe) data reduction in half-year medians and the mean of all medians throughout the patient’s life. We calculated four different IDCs related to age: IDC-A (< 6 years), IDC-B (6-12 years), IDC-C (13-18 years) and IDC-D (> 18 years). To evaluate the fluctuation of Phe values we calculated the standard error of the estimate of the regression of Phe concentration over age. Development quotient was calculated with the Brunet-Lezine test (< 4 years). Intelligence quotient was evaluated with the Kaufman Bit Intelligence Test (K-Bit), Wechsler Intelligence Scale for Children-Revised (WISC-R) and Wechsler Adult Intelligence Scale Third Edition (WAIS III). Results: Cross-sectional study: The IDC in age groups were significantly different and so were the number of patients with good, acceptable and poor IDC related to age (p < 0.001). Sampling frequency was good in 72, acceptable in 23 and poor in 10 patients. The general intelligence (101 ± 10) correlated negatively with the IDC (p < 0.0001) and Phe fluctuations (p < 0.004). Longitudinal study: Significant differences were observed between the IDC through the patients’ lifetime except in the adolescent/adult period. Conclusions: 85% of PKU patients showed good/acceptable quality of dietary control. General intelligence correlates with the IDC at all ages, which highlights the importance of good control to achieve good prognosis.

Keywords

Phenylketonuria, Phenylalanine, Dietary treatment, Intelligence quotient, Recommendations.