IMPACT OF METABOLIC CONTROL IN PHENYLKETONURIC PATIENTS AND ITS RELATIONSHIP WITH NEUROLOGICAL OUTCOMES

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OBJECTIVE AND METHODS
The main objective of this study was to analyze the degree of neurocognitive development deficit in a cohort of patients with PKU through the assessment of their IQs and the influence that plasma phenylalanine levels might exert on this assessment. This cross-sectional study examined 14 patients diagnosed with PKU who performed a K-BIT test. The Kaufman Brief Intelligence Test (K-BIT) is a motor-free screening measure of intelligence that yields Verbal, Nonverbal, and Composite IQ estimates. In addition, mean, median and min/max range of Phe levels for the two years prior to the test and at the time of the test were obtained.

RESULTS
Well-controlled patients at the time of the test (Phe < 600µmol/L) obtained an average result of 50.3; 66.88 and 56.00 points in the K-BIT test (verbal, nonverbal, and composite IQ categories respectively) Vs 26.10; 40.83 and 35.33 in non controlled patients (Fig 1a for composite category).

Regarding the median of Phe levels in the two years prior to the assessment, controlled patients obtained a mean of 43.75; 57.70 and 51.77 Vs 19.00; 43.50 and 23 in non controlled patients (Fig 1b for composite category).

Fig. 1a. Phe levels at the time of the test
Fig. 1b. Median Phe levels 2y prior the test
Statistically significant differences on the results of the K-BIT test measured in percentiles (85 Vs 47.73; p = 0.028) were seen in the nonverbal section and the intake of sapropterin (not in the verbal and composite IQ).

In addition, an inversely proportional correlation was also found between the K-BIT results in its verbal and composite categories with Phe levels at the time of the test (r: -0.553 and -0.556 respectively; p = 0.040 and 0.039).

**CONCLUSIONS**

The results of the K-BIT test seem to depend on the good control of phenylalanine levels in patients with PKU, especially at the time of the test. It seems clear that early diagnosis is vital for the control of phenylalanine levels, which underscores the importance of early diagnosis. Taking sapropterin improves phenylalanine levels and it seems to improve K-BIT test results.