NEUROPSYCHOLOGICAL PROFILE OF PARENTS OF PHENYLKETONURIA PATIENTS

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Introduction

The Phenylketonuria (PKU) is a disorder caused by an inborn error of metabolism. Until now some studies explored cognitive profile of carriers by means of general intelligence tests (i.e. Wechsler intelligence test for adults) and did not investigate specific cognitive functions such as memory, executive functions and visuospatial abilities. The aim of the present study was to explore neuropsychological profile of PKU patients' parents using a comprehensive neuropsychological battery.

Methods

We enrolled 12 parents (7 females and 5 males) of patients affected by PKU and 12 healthy subjects, matched for age, education and sex. All subject underwent standardized neuropsychological tasks for assessment of the following cognitive domains: frontal/executive functions, memory, visuospatial ability. Frontal lobe functions was evaluated by means of the Copy task of Complex Figure of Rey, the Trail Making Test (TMT) and the classic version of the Stroop Color-Word Test. Verbal long-term memory was evaluated by Rey's auditory 15-word learning test; verbal and visuospatial short term memory were assessed by span task and Corsi block tapping task, respectively. Perceptual visuospatial abilities were investigated by means of the Judgment of Line Orientation of Benton, form H.

Results

The two groups did not differ on age, gender and education. There was a statistically significant difference between the two groups on cognitive tests assessing verbal memory and executive/frontal functions. Carriers group performed worse than control group on TMT: part B, TMT: B-A and on interference task of Stroop Test. Moreover, significant differences between the two groups were found on non executive tasks of Stroop Test, i.e. reading and colours denomination tasks. No subjects belonging to both groups achieved pathological scores on select neuropsychological tasks with respect to Italian normative data.

Demographic and cognitive comparisons between heterozygous parents of PKU patients and Healthy controls

	Parents (n=12)	Controls (n=12)	U Test	p
Age (years)	64.8 ± 7.7	63.3 ± 6.1	65.0	0.347
Educational level (years)	6.3 ± 3.6	7.0 ± 3.2	72.5	0.560
RAVLT-immediate	34.8 ± 6.3	44.8 ± 7	23.5	0.001
RAVLT-delayed	7.9 ± 1.9	9.7 ± 2.1	45.0	0.046
Corsi's Test	4.2 ± 1.4	4.5 ± 0.6	72.5	0.515
Verbal Span	3.5 ± 0.5	3.8 ± 0.6	72.0	0.487
TMT-A	89.6 ± 60.4	56.5 ± 42.2	59.0	0.198
TMT-B	267.8 ± 173.	120.2 ± 54.3	25.0	0.002
TMT-B-A	178.1 ± 127.4	71.1 ± 49.9	29.0	0.004
Stroop Test- Word	40.8 ± 19.2	59.2 ± 6	29.0	0.004
Stroop Test-Color	26.6 ± 11.0	40.9 ± 7.8	22.0	0.001
Stroop Test-Interference	10.7 ± 8.0	15.5 ± 4.5	40.5	0.023
BJLOT	15.8 ± 8.8	18.0 ± 4.1	76.0	0.705

RAVLT: Rey Auditory Verbal Learning Test; ROCF: Rey Osterrieth Complex Figure; TMT: Trail Making Test; BJLOT: Benton Judgment Orientation Lines; U Test: Mann-Whitney U Test. Significant difference (after Bonferroni correction; $p<0.005$) between the two groups are shown in bold

Conclusions

The findings evidenced reduced control/executive functions (i.e. set-shifiting and inhibitory control) in PKU parents, when compared to healthy subjects, suggesting a dysfunction of the frontal lobes. Moreover, PKU parents differed significantly from healthy group on non-executive tasks of Stroop Test (i.e. word reading and colour naming tasks); this finding may suggest reduced processing speed.

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