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Neuropsychological profile of children with mitochondrial disorders L Varvogli*1,2 and S Waisbren³

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Background

To describe the neuropsychological profile of children with mitochondrial disorders (myopathies and encephalomyopathies), according to the following questions: (a) do children diagnosed through distinctive clinical symptoms have the same neuropsychological profile as children diagnosed through genetic, histologic, radiologic, or biochemical testing? (b) is there a typical cognitive profile for the mitochondrial disorders in general? (c) is there a typical social/emotional/behavioral profile for the mitochondrial disorders in general? (d) do younger children have a neuropsychological profile that differs from that of older children?

Method

Comparable neuropsychological tests were used for children of different ages, including scales of adaptive behavior, behavioral questionnaires and measures of language, academic, and developmental and cognitive skills. Fiftyseven children, (6-18 years) were assessed. The results were analyzed using SYSTAT programs.

Results

(a) the groups of children diagnosed through different methods had the same neuropsychological profile, (b) characterized by severe deficits in verbal and nonverbal skills, as well as reduced functional skills. Severe mental retardation characterizes over 50% of this sample. (c) There are behavioral and emotional difficulties of anxiety and attention deficit. (d) Younger children had better neuropsychological functioning than older ones.

Discussion

Mitochondrial disorders are associated primarily with sensory-motor difficulties, including gross and fine motor, speech, vision, and hearing difficulties, with compromised developmental and/or cognitive skills. Comprehension, social awareness and reasoning tend to be spared.