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A Rare Form of Rapidly Progressive Parkinsonism Associated with Dementia

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Abstract : Objective: We describe a patient with late onset phenylketonuria which presented with rapidly progressive dementia and parkinsonism that were reversible after management. Background: Phenylketonuria is an autosomal recessive disorder due to mutations in the phenylalanine hydroxlase gene. It normally presents in childhood, in rare cases, however, it may have its onset in adulthood and may mimic other neurological disorders. Case description: A previously normal functioning, 59 year old man was admitted for blurred vision, cognitive impairment and gait difficulty which emerged over the past eight months. In neurological examination he had brisk reflexes, slow gait and left-dominant parkinsonism. Mini-mental state examination score was 25/30, neuropsychological testing revealed a dysexecutive syndrome with constructional apraxia and simultanagnosia. In cranial MRI there were bilateral diffuse hyper-intense lesions in parietal and occipital white matter with no significant atrophy. Electroencephalography showed diffuse slowing with predominance of teta waves. In cerebrospinal fluid examination protein level was slightly elevated (61mg/dL), oligoclonal bands were negative. Electromyography was normal. Routine laboratory examinations for rapidly progressive dementia and parkinsonism were also normal. Serum amino acid levels were determined to explore metabolic leukodystrophies and phenylalanine level was found to be highly elevated (1075 µmol/L) with normal tyrosine (61,20 µmol/L). His cognitive impairment and parkinsonian symptoms improved following three months of phenylalanine restricted diet. Conclusions: Late onset phenylketonuria is a rare, potentially reversible cause of rapidly progressive parkinsonism with dementia. It should be considered in the differential diagnosis of patients with suspicious features.

Keywords : dementia, neurology, Phenylketonuria, rapidly progressive parkinsonism
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