Case Report

A Case of gait disorder due to Combined methylmalonic aciduria and Homocystinuria

abstract

This disorder is too rare that about 100 patients have been reported in the world.In this condition a liver enzyme (methylmalonyl CoA mutase) which should carry out one of many thousands of chemical processes that turn protein into energy or body tissues is defective. Clinical presentation of this disorder in first months of life may be failure to thrive, lethargy, poor feeding, mental retardation, and seizures. Late-onset manifestations include other neurologic findings eg; dementia, myelopathy and gait disorder. Increases in levels of plasma methylemalonic acid and homocystine confirm the diagnosis. Unlike patients with classic homocystinuria, plasma level of methionine is normal in these patients.

The patient was 13.5 years old girl that presented to us due to covulsion, loosing the ability to walk, loosing appetite, developing urinary incontinence and showing intellectual regression. In a number of investigations, there was severe increasing in urinary and plasma levels of methylemalonic acid and homocysitine. The patient has commenced on treatment with high doses of vitamine B_{12}, Betaine and carbamazepine and also physiotherapy. Following the treatment marked improvement in neurologic and mental state appeared and also Methylmalonic acidemia and homocystinuria was controled.

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Key words: Methylmalonic aciduria / Homocystinuria / Convulsion /Betaine