Neuropathology

TETRAPARESIS AS AN INITIAL MANIFESTATION OF BIOTINIDASE DEFICIENCY

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Biotinidase deficiency is an autosomal recessive disorder and causes the deficiency of 4 biotin-containing carboxylases. The prevalence is estimated at 1 in 60 000 births.

Biotinidase deficiency is associated with a wide spectrum of clinical manifestations, including abnormalities of the neurological, dermatological, immunological, and ophthalmological systems.

Spinal cord demyelination as a manifestation of biotinidase deficiency has been infrequently described.

We present a case of 2.5-year-old boy complained of tetraplegia and difficulties in breathing due to Biotinidase deficiency.

Abdominal examination revealed hepatomegaly and splenomegaly.

Also her parents were first-degree cousins. Therefore, tandem mass spectroscopy and urine organic acid analysis were planned to exclude metabolic disorders

Urinary organic acid analysis revealed elevated levels of methyl malonic acid and 3-hydroxy isovaleric acid.

serum biotinidase activity was found to be 3.9 nmol/min/ml. Oral biotin at a dose of 1 mg/kg daily was initiated.

A marked improvement of his neurological deficit was noted over a period of 15 days after treatment and cutaneous manifestations resolved within 3 weeks.

Myelopathy due to Biotinidase deficiency is a challenging diagnosis.

Biotinidase deficiency should be included in the differential diagnosis of children presenting with demyelinating spinal cord disease.