Radiology Case Reports 13 (2018) 592–595

Available online at www.sciencedirect.com

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journal homepage: http://Elsevier.com/locate/radcr

Neuroradiology

Molybdenum cofactor deficiency: Neuroimaging findings

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ARTICLE INFO

Article history:
Received 19 December 2017
Accepted 21 February 2018
Available online

Keywords:
Molybdenum cofactor deficiency
MRI
Spectroscopy

ABSTRACT

Molybdenum cofactor deficiency is an extremely rare and fatal metabolic disorder that should be considered in the differential diagnosis of hypoxic-ischemic encephalopathy. Magnetic resonance imaging findings are useful in diagnosis. The short-echo-time magnetic resonance spectrum was characterized by a total loss of signal and lipid and lactate peaks. In this case, conventional magnetic resonance imaging and magnetic resonance spectroscopy findings of this extremely rare disease whose pathophysiology was not known were presented.

Introduction

Molybdenum cofactor deficiency (MCD) is an extremely rare and fatal metabolic disorder that is characterized by severe and progressive neurologic deterioration in early infancy. Less than 150 cases have been reported in the literature [1]. MCD is an autosomal recessively inherited disorder and reveals findings within the first few days after birth. The major clinical findings are severe neonatal seizures, feeding difficulties, and a progressive neurologic deficit. Diagnosis is late in many cases, and these cases usually pass away in early infancy. Diagnosis can be made by typical magnetic resonance imaging (MRI) findings in the early period. Progressive neurodegeneration is unavoidable in cases with early diagnosis. We presented the clinical presentation and characteristic conventional and spectroscopic MRI findings on this under-recognized disease.

Case

An 11-month-old female patient came to the pediatric neurology clinic with an increase in seizure frequency and serious eating difficulty. She was the first child of second-degree consanguineous parents. There was no history of neurologic diseases in the family. There was a history of three previous abortus of the mother before this delivery. The patient was born to a 41-year-old mother at normal gestational week by vaginal delivery, weighing 3050 g. The patient was diagnosed with infantile spasm-like seizures 1 week after birth, which were controlled

Competing Interests: The authors declare that there are no financial or other relations that could lead to a conflict of interest.

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https://doi.org/10.1016/j.radcr.2018.02.025

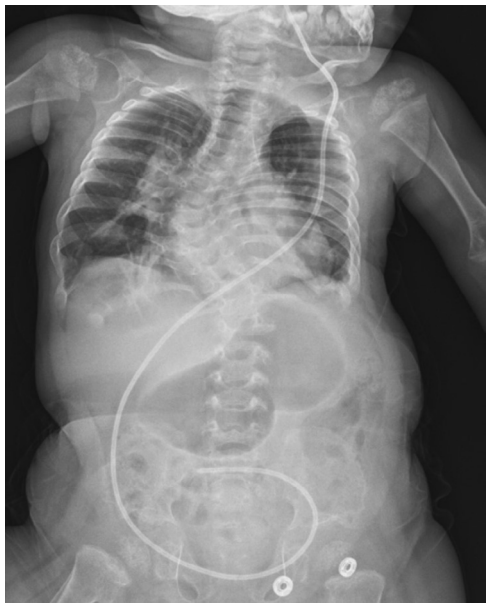

Fig. 1 – On x-ray involving the chest and the abdomen, rotatory scoliosis of the thoracal vertebrae was noted in addition to the ventriculoperitoneal shunt catheter that extended along the cranium and the abdomen.

with phenobarbital. Physical examination revealed microcephaly, hypotonia, and hyper-reflexia. The head circumference was 42.6 cm (between the 3rd and the 10th percentiles), the weight was 7250 g (between the 3rd and the 10th percentiles), and the height was 71 cm (between the 3rd and the 10th percentiles). The measurements were consistent with developmental retardation. Electroencephalographic recordings manifested diffuse, bilateral, and hemispheric epileptiform discharges. There was no detectable uric acid in the urinary sample, indicated as 0 mg/dL. Serum uric acid levels revealed a progressive fall. Serum uric acid measurements obtained intermittently at different times were less than 2 mg/dL on the first and the second measurements, was less than 1 mg/dL on the third measurement, and was 0 mg/dL on the last measurement, which were lower than the normal limits (N: 2.6-6.0 mg/dL). On posterior-anterior chest x-ray, a shunt catheter, which was implanted for hydrocephalus in an external center, was seen, and rotatory scoliosis of the thoracal vertebrae was noted (Fig. 1).

MRI was performed for diagnosis (1.5-T Siemens Magnetom Aera, Germany). MRI of the brain revealed extensive subcortical and periventricular white matter loss, cystic encephalomalacia and hyperintensity in white matter, dysgenesis of the corpus callosum, and ventriculomegaly with generalized volume loss (Fig. 2). In addition, focal polymicrogyria and focal agyria in the cerebral hemisphere at the cortical location,

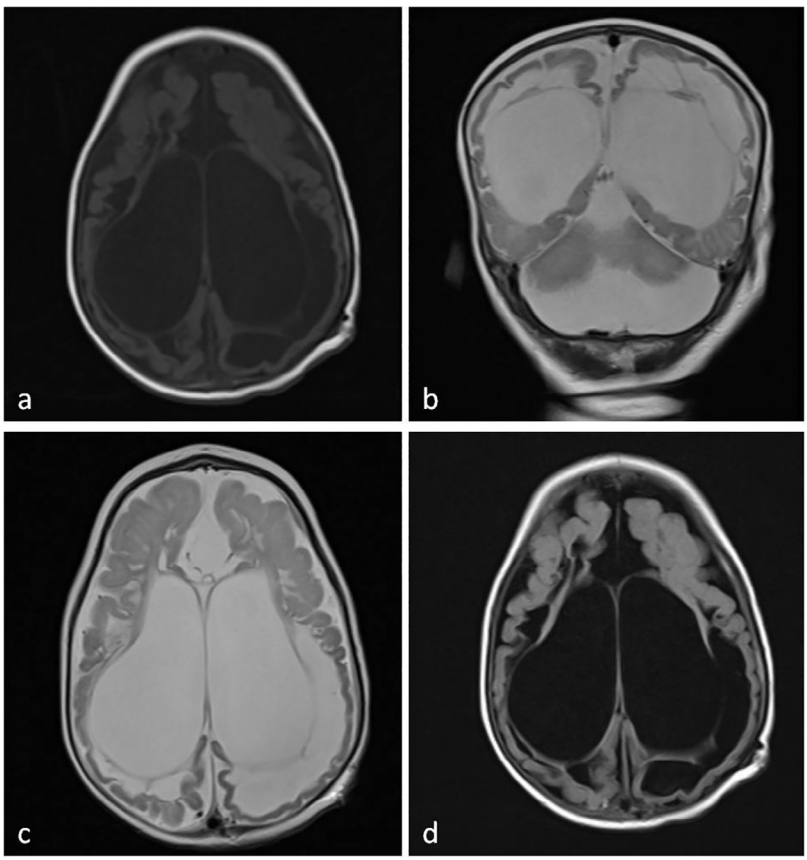

Fig. 2 – T1-weighted axial (A), T2-weighted coronal (B), T2-weighted axial (C), and axial fluid-attenuated inversion recovery sequence (D) of magnetic resonance images show ventriculomegaly, cystic encephalomalacia, and extensive subcortical and periventricular white matter loss and hyperintensity in white matter with atrophy.