Ventricular septation on MRI could be diagnostic for PDHD: Two case reports

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INTRODUCTION

Pyruvate dehydrogenase (PDH) deficiency is a rare metabolic disorder resulted with impairment in the aerobic carbohydrate metabolism and in the use of carbohydrate as the source of oxidative energy. PDH deficiency is commonly associated with lactic acidosis, progressive neuromuscular and neurological degeneration (1). In this review, we aimed to show ventricular septation in magnetic resonance imaging (MRI) as playing a critical role on diagnosis of PDH disease.

CASES

Patient 1, a 43-month-old female patient presented with microcephaly, hypotonia, neuromotor retardation, and infantile spasm seizures that started at 5 months old. Fetal MRI performed with the suspicion of prenatal intracranial bleeding revealed corpus callosum dysgenesis and dilatation in the lateral ventricles. Deep tendon reflexes were hyperactive, limited dorsiflexion in the ankles and the presence of clonus were seen. She had axial hypotonicity. Modified hypsarrhythmia was detected in the EEG. Brain MRI revealed cerebral atrophy due to bilateral posterior frontoparietal and temporal white matter loss, and hemosiderosis with hemorrhage sequelae in the left lateral ventricle and right frontotemporal parenchyma, enlargement of the ventricles and sulci secondary to the loss of ventricles, intraventricular septations, atrophy in the brain stem (Fig 1. A, B). Etiological investigations revealed normal vitamin B12, creatine phosphokinase, and serum amino acids.

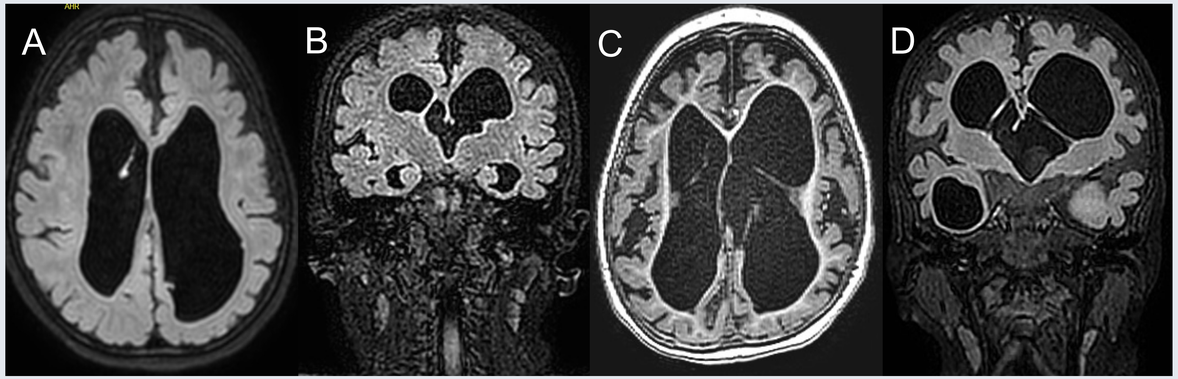

Figure 1. Brain MRI in patient 1 (A-B) and patient 2 (C-D) shows cerebral atrophy, enlargement of the ventricles, and lateral ventricular septa.

Patient 2 presented at age of 4 months with severe hypotonia and seizures. Deep tendon reflexes were hyperactive, axial hypotonicity, spasticity at lower extremities and the presence of clonus were seen. She had epileptic spasms and hypsarrhythmia on EEG. MRI report resulted as cerebral, cerebellar diffuse atrophy, compensated hydrocephalus, septa in the lateral ventricles, hypogenesis of the corpus callosum, arachnoid cyst in the left middle cranial fossa (Fig1 C-D). Clinical features with low energy metabolism and MRI findings with intraventricular septation of both patients directed us to genetic test for metabolic diseases. At patient 1 PDHA1 p.R263* (c.787C>T) heterozygous, Patient 2 PDHA1 p.Asn381fs* (c.1139_1140insAATC) heterozygous mutation detected and ketogenic diet started.

CONCULSIONS

Prenatal neuropathological changes in PDH deficiency consist of a variety of structural lesions could be seen in MRI. Cases may present with cerebral atrophy, complete or partial callosal agenesis, periventricular grey matter heterotopias, or cortical abnormalities, associated with periventricular cysts, white matter abnormalities and intraventricular septations. Prenatal energy failure due to PDH deficiency may account for these malformations**.** The presence of ventricular septations may also be the result of this destructive process (2). Therefore, we aimed to indicate the presence of ventricular septation on MRI with atypical perinatal history is important to consider PDH deficiency in the differential diagnosis as a rare cause.

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