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Case report

Hypoxic ischemic encephalopathy in a case of intranuclear rod myopathy without any prenatal sentinel event

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Abstract

Intranuclear rod myopathy (IRM), a variant of nemaline myopathy, is characterized by the presence of nemaline bodies in myonuclei. We report a case of IRM presenting with hypoxic ischemic encephalopathy (HIE). There were no prenatal complications caused by fetal brain injury. Although no nemaline bodies were observed in the cytoplasm, intranuclear rods were observed in some fibers under light and electron microscopy. Molecular analysis identified a heterozygous variant, c.449C>T (p.Thr150Ile), in *ACTA1*. On magnetic resonance imaging at 9 days of age, injuries to the basal ganglia, thalamus, and brainstem consistent with perinatal HIE were seen. Respiratory insufficiency at birth was strongly suspected to be the cause of HIE. Our case highlights that a patient with a congenital neuromuscular disorder who presents with severe respiratory dysfunction requiring substantial resuscitative efforts at birth can be complicated by HIE without any prenatal sentinel event. Prenatal detection of neuromuscular disorders, careful management of delivery, and neonatal resuscitation and adequate respiratory management are important in preventing irreversible brain injury in these patients.
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Keywords: Nemaline myopathy; Intranuclear rod myopathy; Hypoxic ischemic encephalopathy; Congenital myopathy; Magnetic resonance imaging

1. Introduction

Intranuclear rod myopathy (IRM) is considered a variant of nemaline myopathy (NM) in terms of its pathological features. Patients with IRM present with symptoms similar to those of severe infantile-type NM, but have worse outcomes [1]. We report a case of IRM presenting with hypoxic ischemic encephalopathy (HIE). Cases of congenital neuromuscular disorders with perinatal brain injury complications have so far not been investigated systematically. We have reviewed the literature to investigate the association between HIE and congenital neuromuscular disorders in patients presenting with severe respiratory dysfunction requiring substantial resuscitative efforts at birth.

2. Case report

A 27-year-old woman (gravida 4, para 3) gave birth to a male infant weighing 3060 g by spontaneous

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delivery at a gestational age of 39 weeks. Her family history was unremarkable and her three older children were healthy with no muscle weakness. Although in retrospect she described feeling weak fetal movements as compared to her previous normal pregnancies, there were no apparent maternal or fetal complications, including polyhydramnios, until delivery. Decreased fetal movements and joint contractures were not detected on routine ultrasound. Umbilical cord prolapse, ruptured uterus, and placental abruption were not observed during labor. Fetal heart rate monitoring did not reveal prolonged fetal bradycardia until delivery. At birth, the neonate had dyspnea, cyanosis, and generalized muscle hypotonia. The Apgar scores at 1 min and 5 min were both 2. Umbilical arterial pH was 7.44 and base excess was 4.2 mmol/L. After resuscitation via intubation with adrenaline, the neonate was transferred to our hospital. On admission, he was severely hypotonic, with marked muscle weakness. Anti-gravity movements were not observed, and spontaneous movement was limited only to subtle movements of the extremities. He was not able to suck, and deep tendon reflexes were absent. He had a fracture of the right humerus. Respiratory insufficiency required mechanical ventilation. Arterial gases on admission revealed mixed acidosis with pH 7.27, $PaCO_2$ 50.6 mmHg and base excess −5.7 mmol/L. Serum lactate on admission was elevated to 8.1 mmol/L. Serum creatine kinase was elevated to 2233 U/L on day 2, with a repeat measurement of 133 U/L. The electroencephalography showed depressed background activity and ictal discharges.

Brain magnetic resonance imaging (MRI) was performed at 9 days of age (Fig. 1). T1-weighted images showed hyperintensity of the lateral thalamus, posterior putamen, globus pallidus and dorsal midbrain. T2-weighted images showed hyperintensity of the posterior globus pallidus and dorsal midbrain. White matter injury was not found. These findings were consistent with a form of brain injury previously termed "total asphyxia" (Myers) [2] or "profound asphyxia" (Barkovich) [3], which is seen in term infants with HIE involving abrupt profound anoxia.

Biopsy of the biceps brachii muscle demonstrated that most fibers were very small, as revealed by hematoxylin and eosin staining. The adenosine triphosphatase staining revealed type 2 fiber predominance. Intranuclear rods were observed in some fibers under light and electron microscopy (Fig. 2), leading to a pathological

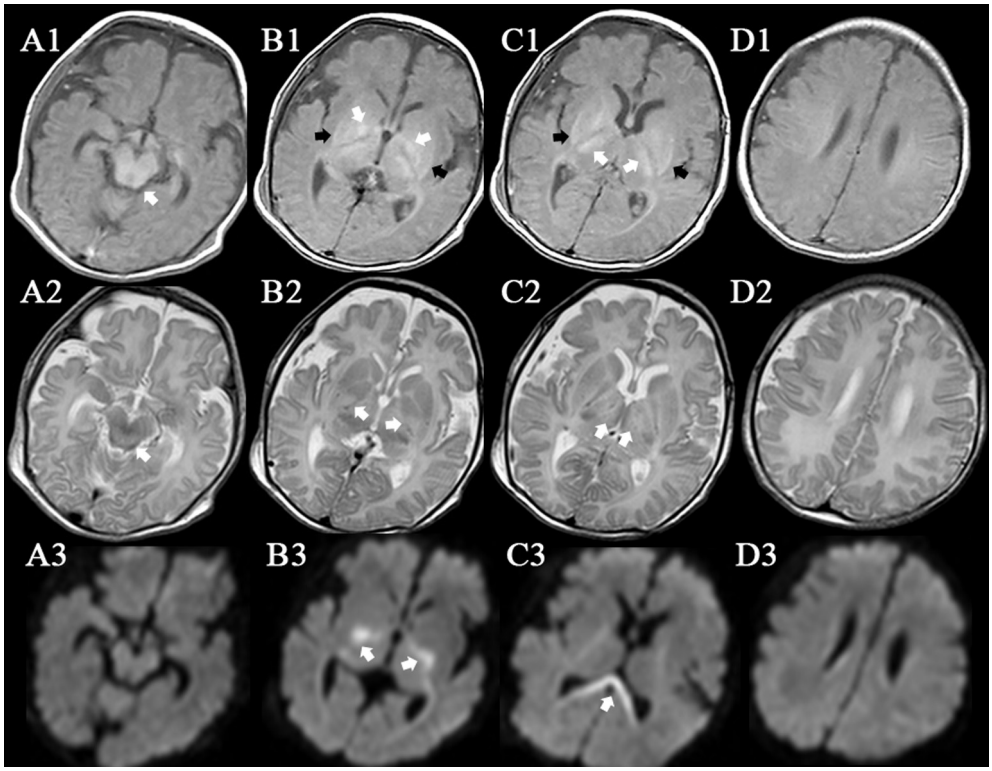

Fig. 1. Brain magnetic resonance imaging. T1-weighted images (A1, B1, C1, D1) showed hyperintensity of the dorsal midbrain (A1: white arrow), globus pallidus (B1: white arrow), posterior putamen (B1, C1: black arrow), and lateral thalamus (C1: white arrow). Hyperintensity of the internal capsule was not observed. T2-weighted images (A2, B2, C2, D2) showed hyperintensity of the dorsal midbrain (A2: white arrow), posterior globus pallidus (B2: white arrow), and central part of thalamus (C2: white arrow). Diffusion-weighted images (A3, B3, C3, D3) showed hyperintensity of the globus pallidus (B3: white arrow) and splenium of the corpus callosum (C3: white arrow). White matter injury was not present. Cortical highlighting was not observed.

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