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Case report

Bilateral congenital lumbar hernias in a patient with central core disease – A case report

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Abstract

Congenital lumbar hernias are rare malformations caused by defects in the development of the posterior abdominal wall. A known association exists with lumbocostovertebral syndrome; however other associated anomalies, including one case with arthrogryposis, have been previously reported. We present an infant girl with bilateral congenital lumbar hernias, multiple joint contractures, decreased muscle bulk and symptoms of malignant hyperthermia. Molecular testing revealed an R4861C mutation in the ryanodine receptor 1 (*RYR1*) gene, known to be associated with central core disease. This is the first reported case of the co-occurrence of congenital lumbar hernias and central core disease. We hypothesize that ryanodine receptor 1 mutations may interrupt muscle differentiation and development. Further, this case suggests an expansion of the ryanodine receptor 1-related myopathy phenotype to include congenital lumbar hernias.

Keywords: Congenital lumbar hernia; Central core disease; Malignant hyperthermia; *RYR1*; Arthrogryposis

1. Introduction

Congenital lumbar hernias are rare malformations, with fewer than 80 cases reported worldwide [1]. Most are unilateral [2]. The most common associated malformations are those which make up the lumbocostovertebral syndrome. Lumbocostovertebral syndrome describes an association of congenital lumbar hernias, along with genitourinary, vertebral and rib anomalies. This syndrome is felt to result from a defect in the first lumbar myotome, leading to hypoplasia of the transversus abdominis and oblique muscles [1].

Congenital lumbar hernias have also been associated with a wide-spectrum of other anomalies including VACTERL (vertebral, anal atresia, cardiac defects, tracheoesophageal, renal anomalies, limb defects) association, congenital diaphragmatic hernia, atrial septal defect, focal nodular hyperplasia of the liver, hydrocephalus and absent kidney [3–6]. One case of clubfoot and arthrogryposis has also been reported, suggesting a possible link between congenital lumbar hernias and an underlying neuromuscular disorder [5].

We present the first case of bilateral congenital lumbar hernias in an infant with central core disease; the association suggests that *RYR1* mutations may be the cause of this congenital malformation. Consent was obtained from the patient's family for publication of this case.

2. Case report

The patient was of Northern European descent, and from a non-consanguineous family. Family history was unremarkable. There were normal prenatal ultrasounds and no evidence of polyhydramnios; however decreased fetal movements were noted in the third trimester. The patient was born at term with a birth weight of 3490 g. Bilateral flank swelling were noted shortly

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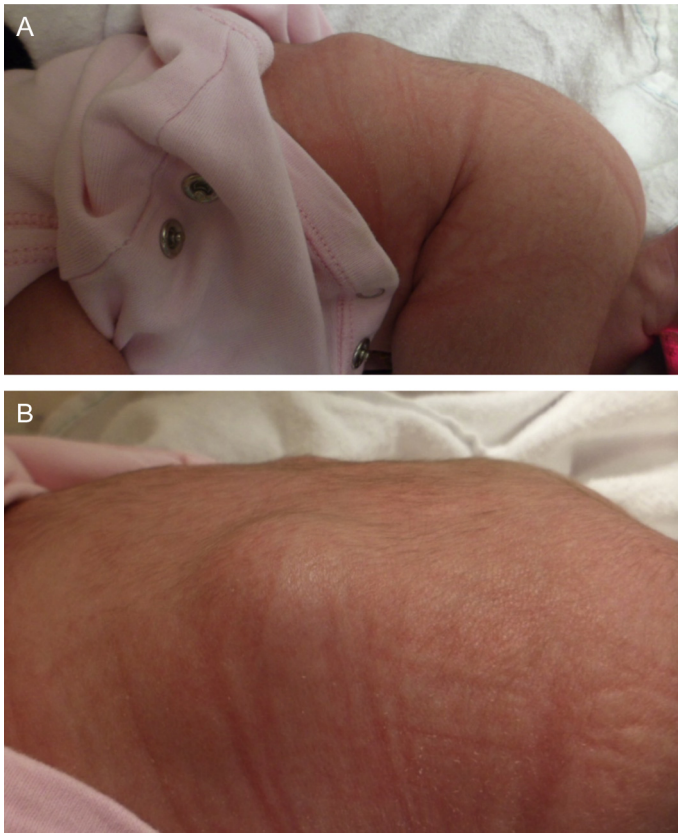

Fig. 1. The patient in the newborn period. Dorsal lumbar hernias are seen containing kidney and bowel.

after birth, and determined to be congenital lumbar hernias on ultrasound (Fig. 1). Multiple joint contractures (arthrogryposis) were evident within the first few days of life, including abnormal finger positioning with adducted thumbs, hyperextension of the proximal interphalangeal joints and flexion of the distal interphalangeal joints, bilateral congenital hip dislocations, and joint contractures at the shoulders, elbows, and knees, and ankles. The patient was noted to have decreased muscle bulk, hypotonia, and generalized muscle weakness, but with preserved anti-gravity movements in both legs. Extraocular movements were preserved. There were no genitourinary anomalies. She did not have any vertebral or rib anomalies on imaging. The neonatal course was further complicated by an ongoing oxygen requirement. The patient was discharged home at 25 days of age off oxygen therapy.

Initial investigations included a creatine kinase which was elevated at 3813 U/L (normal <170) on day one of life, however this decreased to 192 U/L by 1 week and has been normal (≤50 U/L) since. Initial genetic investigations including a newborn metabolic screen, array comparative genomic hybridization (CytoSure ISCA 8x60K v2.0, Oxford Gene technology), and testing for spinal muscular atrophy (Molecular Diagnostic Laboratory, Calgary, Alberta) were normal. An abdominal ultrasound done on day 1 of life revealed lumbar hernias with structurally normal underlying kidneys. Magnetic resonance imaging of abdomen and pelvis done at one month of age showed decreased muscle bulk with increased T1 signal involving

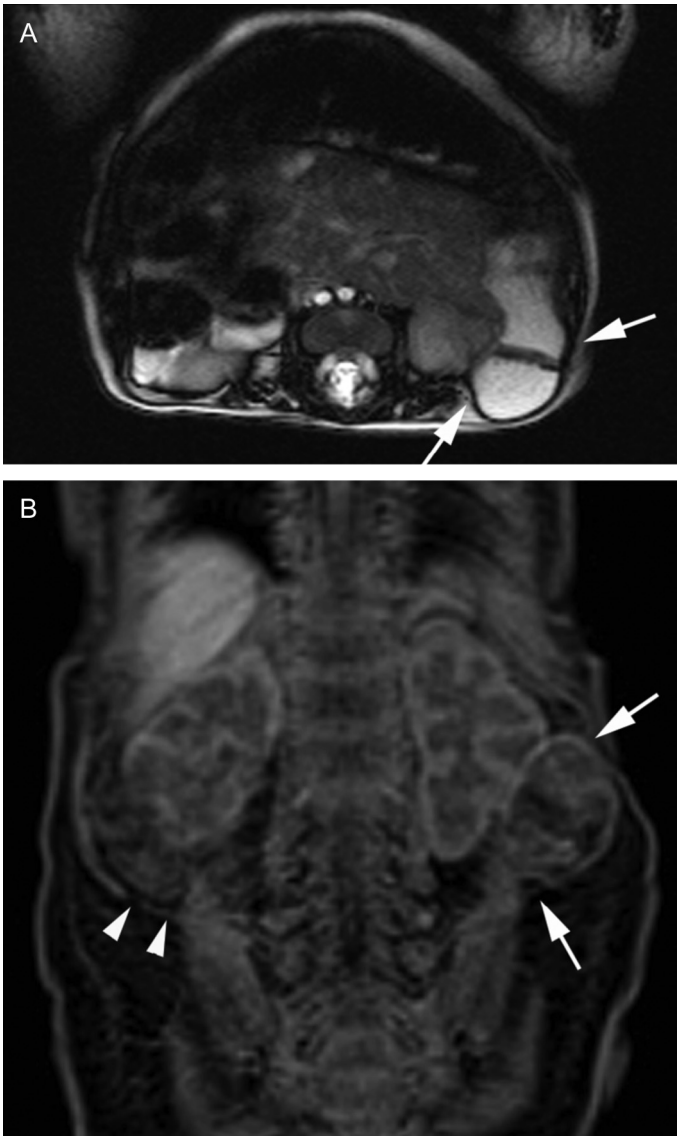

Fig. 2. Lumbar hernia on MRI at age of 5 weeks. Axial T2-weighted image (A) and axial reformated T1-weighed image (B) of abdomen revealed herniation of a fluid-containing bowel loop through the left posterolateral abdominal wall (arrows in A and B), probably via the inferior triangle. Note the normal lower lumbar triangle on the right side (arrow heads in B). Also note the diffusely decreased muscle bulk in abdominal wall and paraspinal regions. Herniated kidneys are not captured in this image.

all the visualized muscles, but most obvious in the larger muscles including the psoas, quadratus lumborum, erector spinae, and latissumus muscles. Herniation of the colon and kidneys through the posterolateral abdominal wall was seen (Fig. 2). Magnetic resonance imaging of the limbs was not performed. Magnetic resonance imaging of the brain done at 4 months of age was normal. An electromyogram done at 4 months showed reduced motor amplitudes consistent with a myopathic process. Electrocardiography was normal. Spine X-rays at birth did not show any vertebral anomalies apart from a mild mid-thoracic scoliosis.

The patient required multiple hospital admissions in early infancy for respiratory infections, failure to thrive, and hypotonia.