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Whole-body muscle MRI characteristics of LAMA2-related congenital muscular dystrophy children: An emerging pattern

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Abstract

Merosin-deficient or LAMA2-related congenital muscular dystrophy (CMD) belongs to a group of muscle diseases with an overlapping diagnostic spectrum. MRI plays an important role in the diagnosis and disease-tracking of muscle diseases. Whole-body MRI is ideal for describing patterns of muscle involvement. We intended to analyze the pattern of muscle involvement in merosin-deficient CMD children employing whole-body muscle MRI. Ten children with merosin-deficient CMD underwent whole-body muscle MRI. Eight of which were genetically-confirmed. We used a control group of other hereditary muscle diseases, which included 13 children (mean age was 13 SD +/- 5.5 years), (8 boys and 5 girls) for comparative analysis. Overall, 37 muscles were graded for fatty infiltration using Mercuri scale modified by Fischer et al. The results showed a fairly consistent pattern of muscle fatty infiltration in index group, which differs from that in control group. There was a statistically significant difference between the two groups in regard to the fatty infiltration of the neck, serratus anterior, intercostal, rotator cuff, deltoid, triceps, forearm, gluteus maximus, gluteus medius, gastrocnemius and soleus muscles. Additionally, the results showed relative sparing of the brachialis, biceps brachii, gracilis, sartorius, semitendinosus and extensor muscles of the ankle in index group, and specific texture abnormalities in other muscles. There is evidence to suggest that whole-body muscle MRI can become a useful contributor to the differential diagnosis of children with merosin deficient CMD. The presence of a fairly characteristic pattern of involvement was demonstrated. MRI findings should be interpreted in view of the clinical and molecular context to improve diagnostic accuracy.

Keywords: Congenital muscular dystrophy type 1; Genetic muscular diseases; LAMA2 gene; Laminin Alpha-2 congenital muscular dystrophy; Merosin-deficient congenital muscular dystrophy.

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