Calf Muscles Hypertrophy in Sarcoglycanopathy

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ABSTRACT

Though calf muscle hypertrophy is thought to be a signatory finding of dystrophinopathies, it can also be observed in other muscular dystrophies. Failure to recognise this may result in diagnostic errors. We present a patient with delta sarcoglycanopathy who had hypertrophy of the brachioradialis, gastrocnemius and extensor digitorum brevis.

Keywords: Calf hypertrophy, dystrophinopathy, sarcogly-canopathy.

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INTRODUCTION

A 15-year-old boy with normal development milestones presented with proximal lower limb weakness for 4 years. There was no family history of any muscle disease. On examination, in addition to proximal weakness, he was found to have hypertrophy of brachioradialis, gastrocnemius, and extensor digitorum brevis muscles bilaterally (Figs 1A to C). Total creatine kinase was elevated (4798), electromyography was myopathic, and immunohistochemistry of muscle biopsy revealed absence of delta-sarcoglycan staining confirming diagnosis of delta sarcoglycanopathy or limb girdle muscle dystrophy 2F (Figs 2A to J).

Dystrophinopathy and sarcoglycanopathy can be very similar, both phenotypically and in clinical course.^{1,2} While calf pseudohypertrophy is conventionally thought

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Figs 1A to C: (A) Bilateral hypertrophied brachioradialis; (B) bilateral hypertrophied calf muscles; and (C) bilateral hypertrophied extensor digitorum brevis

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