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PHOTO ASSAY

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, sarcoglycanopathy.

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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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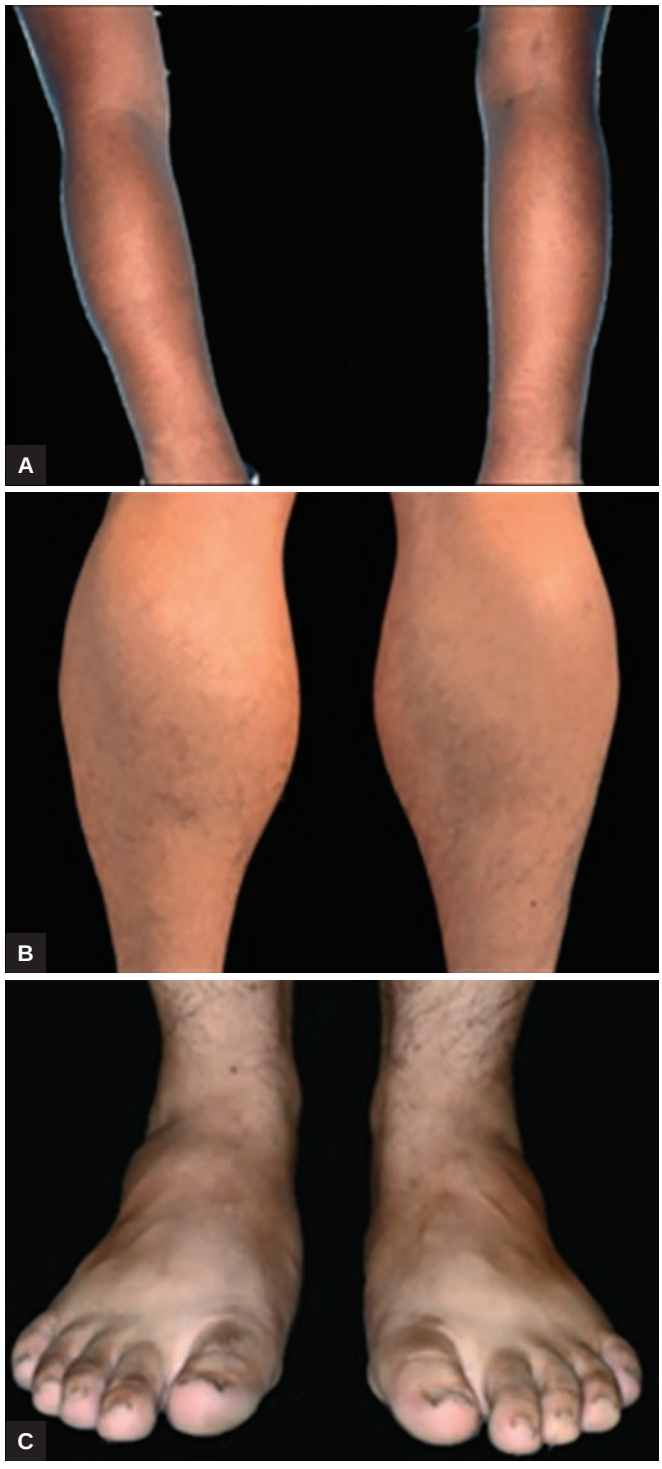
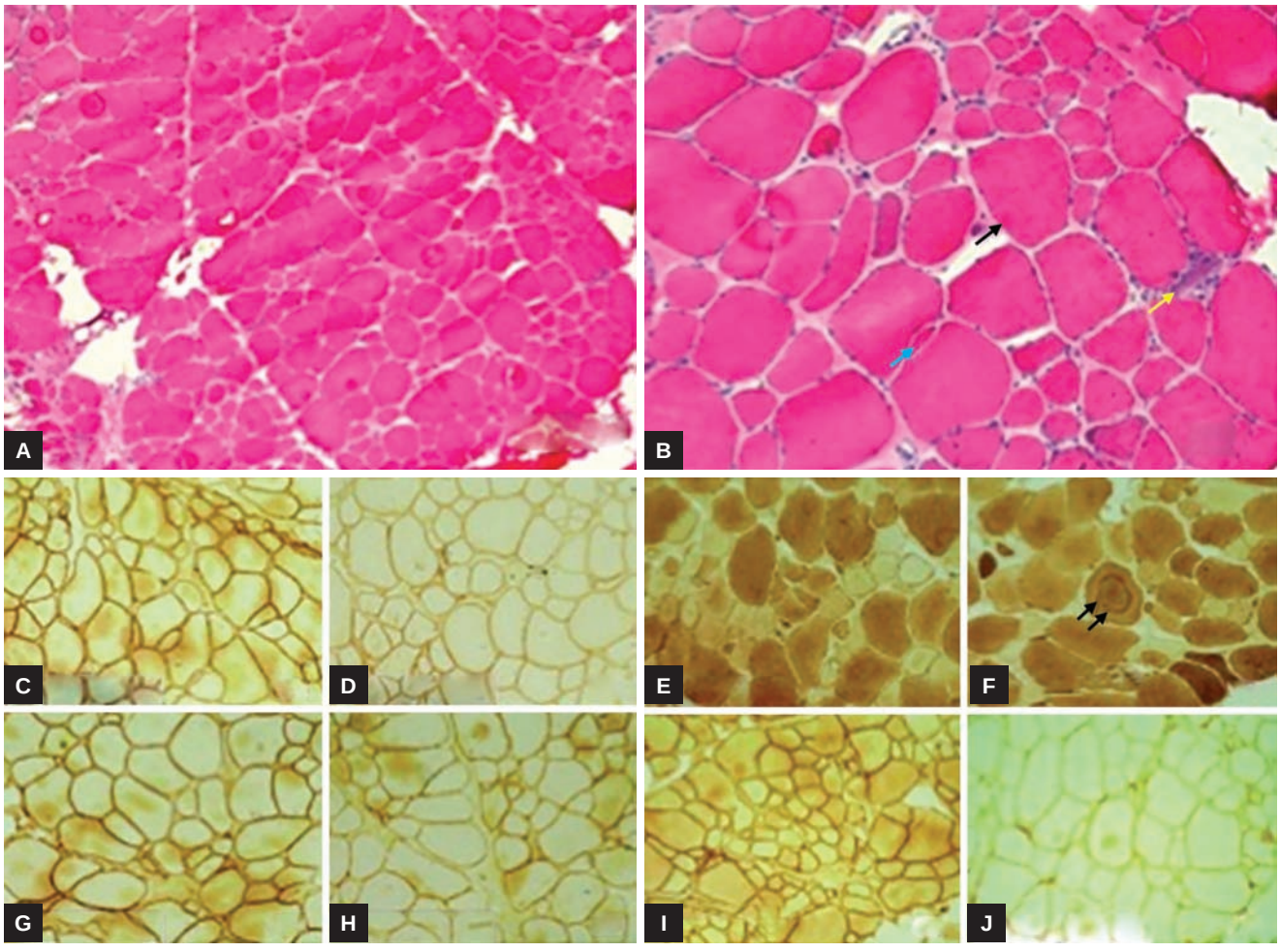


Figs 1A to C: (A) Bilateral hypertrophied brachioradialis; (B) bilateral hypertrophied calf muscles; and (C) bilateral hypertrophied extensor digitorum brevis



Figs 2A to J: (A and B) Hematoxylin and eosin (H&E) photomicrographs showing marked variation in muscle fiber size and shape with mild increase in endomysial connective tissue. The size variation is dominantly due to hypertrophic fibers (black arrow) alternating with atrophic fibers (blue arrows). Occasional basophilic fibers are noted (yellow arrow); (C to J) immunostains for dystrophin (A), dystrophin (B), dysferlin (Hamlet)—1, dysferlin (Hamlet)—2, sarcoglycan alpha, sarcoglycan beta, and sarcoglycan gamma respectively. All show normal staining pattern and intensity. Dysferlin—2 in particular showed abnormal targetoid sarcoplasmic staining within a few muscle fibers (black arrows). Immunostain for sarcoglycan delta showed normal pattern but marked reduction in staining intensity

to be more common in dystrophinopathies,[3] our case highlights the importance of considering sarcoglycanopathy even in the presence of calf muscles hypertrophy.

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