A rare case of congenital fibre type disproportion causing delayed motor milestones

R Waduge[1], N V I Ratnatunga[1], A Padeniya[2], D N K Bowala[1], S Chandraprabha[1]

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Introduction

Congenital myopathies are a clinically, genetically and pathologically heterogeneous group of muscle disorders. Congenital fiber type disproportion (CFTD) is a rare type of congenital myopathy which presents with hypotonia, delayed motor milestones and dysmorphic facies [1, 2].

Case report

A two year and four month old boy who was the second child of a non-consanguineous marriage was investigated for delayed motor mile stones and dysarthria. The patient had an uneventful antenatal history. He started walking at the age of two years. There was no progressive proximal muscle weakness. Examination showed long facies and a squint. Muscle tone in both upper and lower limbs was mildly reduced. Creatine phosphokinase levels were normal, 65U/L (24-195U/L). The nerve conduction study excluded spinal muscular atrophy.

A muscle biopsy of vastus lateralis was done. Enzyme histochemistry for succinic dehydrogenase showed a contrasting size difference in type 1 and 2 fibres. The mean fibre diameter of type 1 and 2 fibres was 6µ and 10µ respectively. The age and sex matched norm of mean fibre diameter is 10-18µ for both type 1 and 2 fibres. The type 1 fibres occupied 64% of fibre population and type 2 fibres 36% approximately (Figures1 and 2). No fibre necrosis, interstitial fibrosis or any other histological abnormalities were detected. Therefore a diagnosis of CFTD was made.

Discussion

The pathological criterion for determining CFTD is the predominance of type 1 fibres that are at least 12% smaller in diameter than type 2 fibres in the absence of any other pathological features [1]. In this case, the difference of mean fibre diameter of type 1 and type 2 fibres was 40%. The ratio of type 1 and 2 fibres was 2:1 (normal ratio is 1:2) with a predominance of type 1fibres. The degree of weakness varies and some show involvement of respiratory muscles. The disease can become static or improvement can occur [1].

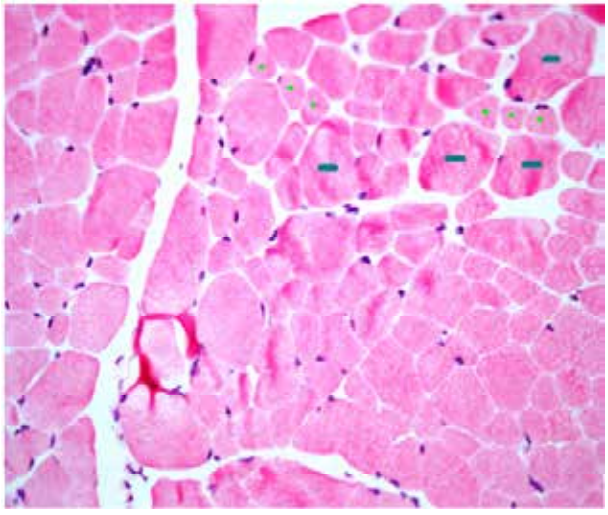

Figure 1. **H & E stain to visualise small fibres with and normal fibers with (x400)**

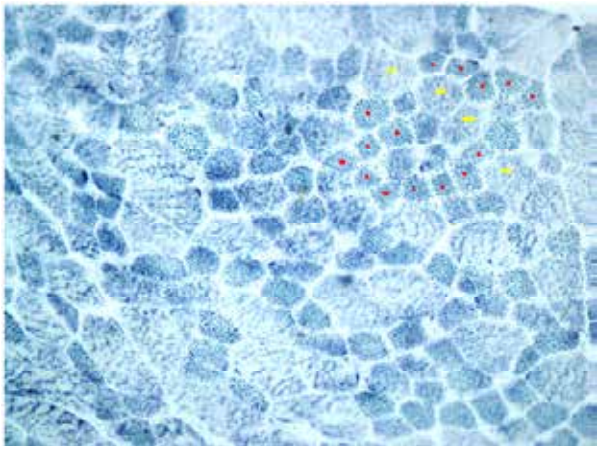

Figure 2. **Succinic dehydrogenase enzyme stain to visualise darkly stained small type 1 fibres with and lightly stained type 2 fibres with (x400)**

[1]*Department of Pathology, Faculty of Medicine, University of Peradeniya and* [2]*Sirimavo Bandaranaike Specialized Childrens Hospital, Peradeniya, Sri Lanka.*

Correspondence: RW, *e-mail: <roshitha2006@yahoo.com>.* *Received 29 August and revised version accepted 29 December 2014. Competing interests: none declared.*

Declaration of Interest

There are no conflicts of interest.

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