**Format:** Abstract


**Congenital muscular dystrophy with inflammation: Diagnostic considerations.**

Konkay K, Kannan MA, Lingappa L, Uppin MS, Challa S.

**Author information**

**Abstract**

**BACKGROUND AND PURPOSE:** Muscle biopsy features of congenital muscular dystrophies (CMD) vary from usual dystrophic picture to normal or nonspecific myopathic picture or prominent fibrosis or striking inflammatory infiltrate, which may lead to diagnostic errors. A series of patients of CMD with significant inflammatory infiltrates on muscle biopsy were correlated with laminin α2 deficiency on immunohistochemistry (IHC).

**MATERIAL AND METHODS:** Cryostat sections of muscle biopsies from the patients diagnosed as CMD on clinical and muscle biopsy features from 1996 to 2014 were reviewed with hematoxylin and eosin (H&E), enzyme and immunohistochemistry (IHC) with laminin α2. Muscle biopsies with inflammatory infiltrate were correlated with laminin α2 deficiency.

**RESULTS:** There were 65 patients of CMD, with inflammation on muscle biopsy in 16. IHC with laminin α2 was available in nine patients, of which six showed complete absence along sarcolemma (five presented with floppy infant syndrome and one with delayed motor milestones) and three showed discontinuous, and less intense staining.

**CONCLUSIONS:** CMD show variable degrees of inflammation on muscle biopsy. A diagnosis of laminin α2 deficient CMD should be considered in patients of muscular dystrophy with inflammation, in children with hypotonia/delayed motor milestones.

**KEYWORDS:** Congenital muscular dystrophies (CMDs); inflammation; laminin α2 (LAMA2); merosin

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