Collagen XII myopathy with rectus femoris atrophy and collagen XII retention in fibroblasts.

Witting N¹, Krag T¹, Werlauff U², Duno M³, Oestergaard ST¹, Dahlqvist JR¹, Vissing J¹.

INTRODUCTION: Mutation in the collagen XII gene (COL12A1) was recently reported to induce Bethlem myopathy. We describe a family affected by collagen XII-related myopathy in 3 generations.

METHODS: Systematic interview, clinical examination, skin biopsies and MRI muscle.

RESULTS: The phenotype was characterized by neonatal hypotonia, contractures and delayed motor development followed by resolution of contractures and a motor performance limited by reduced endurance. DNA analyses revealed a novel donor splice-site mutation in COL12A1 (c.8100 + 2T>C), which segregated with clinical affection and abnormal collagen XII retention in fibroblasts. MRI disclosed a selective wasting of the rectus femoris muscle.

DISCUSSION: COL12A1 mutations should be considered in patients with a mild Bethlem phenotype who present with selective wasting of the rectus femoris, absence of the outside-in phenomenon on MRI, and abnormal collagen XII retention in fibroblasts. This article is protected by copyright. All rights reserved.

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KEYWORDS: Bethlem myopathy; Collagen 12A1 mutation; MRI muscle; clinical phenotype; congenital muscular dystrophy; fibroblast stain

PMID: 29342313 DOI: 10.1002/mus.26067