Bethlem Myopathy Phenotypes and Follow Up: Description of 8 Patients at the Mildest End of the Spectrum.

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Abstract
The classical phenotypes of collagen VI-associated myopathies are well described. Little is known, however, about the progression of patients at the mildest end of the clinical spectrum. In this report, we describe the clinical findings and the results of MRI, muscle biopsy, collagen VI expression in cultured skin fibroblasts and genetic tests of a series of patients with Bethlem myopathy. Our series highlights the existence of mild presentations of this disorder that progresses only slightly and can easily be overlooked. Analysis of the genetic studies suggests that missense mutations can be associated to a milder clinical presentation. Muscle MRI is extremely useful as it shows a pathognomonic pattern in most patients, especially those with some degree of muscle weakness.

KEYWORDS: Bethlem disease; Ulrich disease; collagen VI deficiency; muscle MRI; muscle dystrophy

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