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# Collagen VI involvement in Ullrich syndrome: a clinical, genetic, and immunohistochemical study.

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## Abstract

**BACKGROUND: Ullrich** congenital muscular dystrophy (UCMD) is a form of merosin-positive congenital muscular dystrophy characterized by proximal contractures, distal laxity, rigidity of the spine, and respiratory complications. Recently, a deficiency of **collagen VI** on muscle and skin biopsy together with recessive mutations in the **collagen** 6A2 gene were reported in three families with UCMD. However, the **clinical** spectrum, frequency, and level of heterogeneity of this disorder are not known.

**SUBJECTS AND METHODS:** The authors studied 15 patients (aged 3 to 23.6 years) with a **clinical** diagnosis of UCMD. Linkage analysis to the three **collagen VI** genes was performed in all informative families (n = 7), whereas **immunohistochemical** analysis of **collagen VI** expression in muscle was performed in the remaining cases.

**RESULTS:** An immunocytochemical reduction of **collagen VI** was observed in six patients. Three of the six patients belonged to informative families, and haplotype analysis clearly suggested linkage to the COL6A1/2 locus in two cases and to the COL6A3 loci in the third case. In the remaining nine patients, primary **collagen VI involvement** was excluded based on either the linkage analysis (four families) or considered unlikely based on normal immunolabeling of **collagen VI**. Age and presentation at onset, the distribution and severity of weakness and contractures, and the frequency of nonambulant patients were similar in the patients with and without **collagen VI involvement**. Distal laxity, rigidity of the spine, scoliosis, failure to thrive, and early and severe respiratory impairment were found in all patients by the end of the first decade of life, irrespective of

their maximum motor functional ability or their **collagen** status.

**CONCLUSIONS:** These results suggest that **collagen VI involvement** is relatively common in UCMD (40%); however, the role of this molecule was excluded in a number of cases, suggesting **genetic** heterogeneity of this condition.

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