Characteristic phenotype of Ullrich congenital muscular dystrophy

A 21-year-old woman presented with clinically classic signs of Ullrich congenital muscular dystrophy (figure). Genetic testing of collagen VI genes revealed a homozygous mutation c.2329T>C, p.Cys777Arg in the COL6A2 gene, consistent with the clinical diagnosis.

Collagen type VI–related disorders represent a spectrum of overlapping phenotypes: Bethlem myopathy at the milder end, and Ullrich congenital muscular dystrophy at the severe end. Its clinical features may resemble Emery-Dreifuss muscular dystrophy, but absence of cardiac abnormalities is helpful in distinguishing these 2 disorders. Recognition of typical clinical features can aid in the diagnosis and help to shorten a potentially lengthy diagnostic workup.

AUTHOR CONTRIBUTIONS
Wendy Liew: drafting and revision of manuscript. Basil Darras: interpretation of data and manuscript revision.

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REFERENCES
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