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Teaching Neuro*Images*: Characteristic phenotype of Ullrich congenital muscular dystrophy

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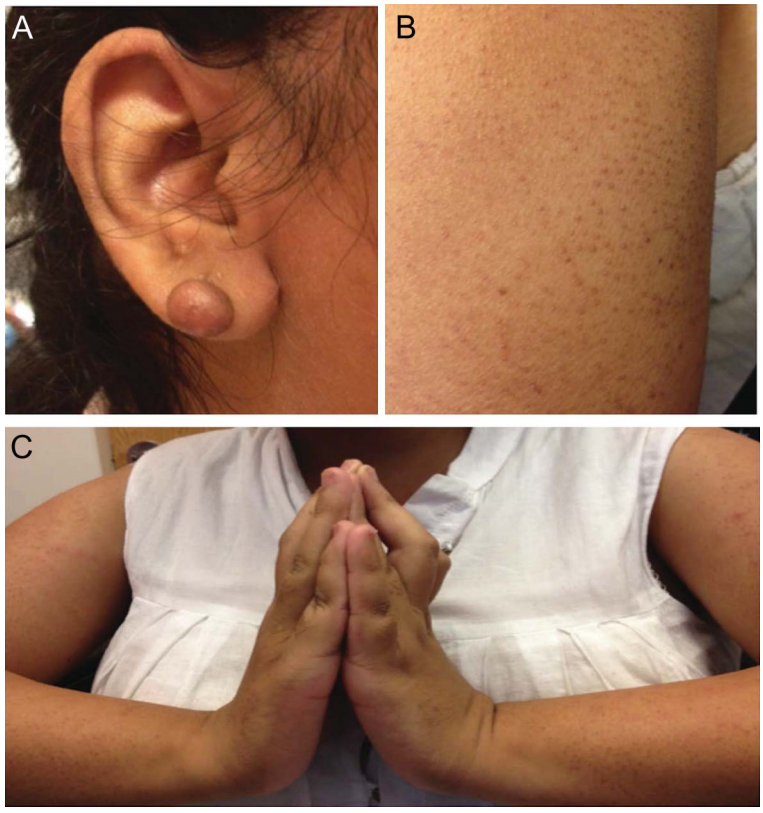
Figure Classic signs of Ullrich congenital muscular dystrophy



A 21-year-old woman with progressive limb-girdle weakness, elbow contractures, and hyperlaxity of distal joints also exhibited the following: (A) keloid formation after ear piercing; (B) follicular hyperkeratosis of the arm; and (C) Bethlem sign—flexion contractures of the fingers on wrist extension.

A 21-year-old woman presented with clinically classic signs of Ullrich congenital muscular dystrophy[1] (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.[2] 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.

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

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