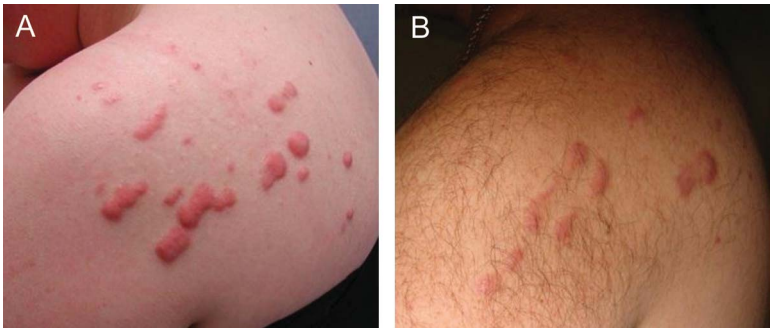
Spontaneous keloid formation in patients with Bethlem myopathy

Figure Spontaneous keloid formation in patients with Bethlem myopathy



(A) A 32-year-old woman who has difficulty arising from a chair, joint contractures, and respiratory insufficiency. She also developed a keloid on her chest. *COL6A3* mutation: exon 16, c.G6517T, p.Gly2053Cys. (B) A 50-year-old man who is dependent on a cane for walking. *COL6A2* mutation: exon 25, c.G1861A, p.Asp621Asn.

A 32-year-old woman and a 50-year-old man with clinically typical Bethlem myopathy developed seemingly spontaneous keloids on their shoulder region (figure). The patients did not recall any significant trauma to the skin of this region.

Bethlem myopathy (MIM #158810) is caused by dominant and recessive mutations in the collagen VI genes: *COL6A1*, *COL6A2*, and *COL6A3*. Skin manifestations include hyperkeratosis pilaris and keloid or "cigarette paper" scar formation.[1,2] Spontaneous keloid formation has not been previously reported. These cases illustrate the importance of considering collagen VI gene mutations in patients with weakness associated with such characteristic skin findings and joint contractures.

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