Skin Biopsy for Diagnosis of Ullrich Congenital Muscular Dystrophy: An Observational Study.

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Abstract
The gold standard diagnostic test for Ullrich congenital muscular dystrophy (UCMD) is molecular testing for COL6 mutation. The facility for genetic testing is sparingly available and it is usually diagnosed by muscle biopsy. The latter is an invasive procedure requiring expertise and sedation. Skin biopsy has shown promise as a simpler diagnostic modality. Eleven and 7 cases, respectively, of phenotypically suspected Ullrich congenital muscular dystrophy and dystrophinopathy underwent simultaneous skin and muscle biopsies, which were subjected to hematoxylin and eosin (H&E) and immunohistochemistry staining for collagen VI and dystrophin 1, 2, and 3. Of the 8 and 5 muscle biopsy-confirmed cases of Ullrich congenital muscular dystrophy and dystrophinopathy, 6 Ullrich congenital muscular dystrophy and 5 dystrophinopathy cases showed absent and preserved COL6 staining, respectively, in the skin biopsy. Skin biopsy as a diagnostic option has shown encouraging results in Ullrich congenital muscular dystrophy. These should be evaluated in larger studies.

KEYWORDS: COL6; UCMD; dystrophinopathy; muscle biopsy; skin biopsy

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