

Classic Physical Exam Findings in Ehlers-Danlos Syndrome

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A 15-year-old man presented with 14 years of skin hyperextensibility, atrophic cigarette-paper scars, easy bruising, and joint hypermobility (Fig. 1a–d), findings consistent with Classical Ehlers-Danlos syndrome (CEDS).

CEDS is an autosomal dominant disorder secondary to COL5A1/COL5A2 gene mutations which encode type V collagen.^{1,2}

Diagnosis is based upon the presence of major criteria (skin hyperextensibility; widened atrophic cigarette-paper scars with poor wound healing; and joint hypermobility)

along with any minor criterion (soft doughy skin, easy bruising, fragile skin, molluscoid pseudotumors, subcutaneous spheroids, joint hypermobility complications, epicanthal folds, hernias, and positive family history), or by the presence of the major skin criterion and either generalized joint hypermobility and/or three or more minor criteria.^{1–3}

Minor trauma leads the dermis to separate, creating skin hyperextensibility, defined by the capacity to stretch skin until feeling resistance for 4 cm or more at a neutral site (neck or ventral aspect of the forearm).⁴ As a result, wound healing is poor, with stretching and widening scars (cigarette-paper-like appearance).² Joint hypermobility is secondary to an increased distensibility of ligaments and joint capsules due to collagen and extracellular matrix alterations.^{5,6}

Knowing and recognizing EDS clinical findings are essential for diagnosing and preventing complications.

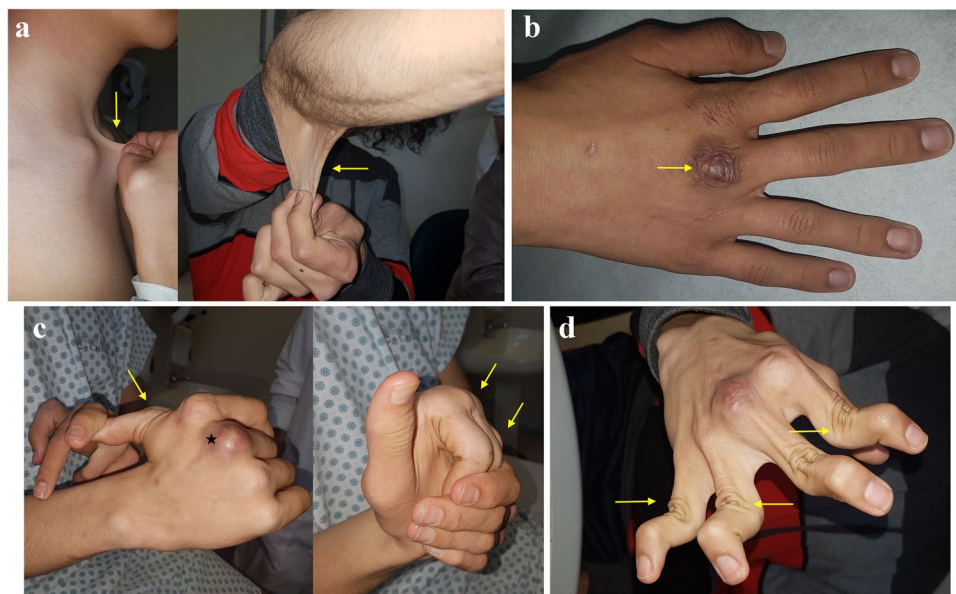


Figure 1 Panel a; skin hyperextensibility (yellow arrow) of the anterior neck and the right elbow. Panel b; Atrophic and widened scar with the appearance of cigarette paper (yellow arrow) on the knuckle of the middle finger of the right hand. Panel c; Joint hypermobility (yellow arrow) of the right hand's metacarpophalangeal joints. Note the atrophic cigarette-paper-like scar on the knuckle of the middle finger of the right hand (black star). Panel d; Joint hypermobility of the interphalangeal joints of the right hand (yellow arrow).

This Study was conducted at the Department of Dermatology, Facultad de Medicina, Pontificia Universidad Católica de Chile.

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Data Availability The data that support the findings of this study are available from the corresponding author, (CVK), upon reasonable request.

Declarations

Conflict of Interest The authors have no conflict of interest to declare.

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