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Ehlers-Danlos Syndrome, Hypermobility Type: An Underdiagnosed Hereditary Connective Tissue Disorder with Mucocutaneous, Articular, and Systemic Manifestations

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Additional article information

Abstract

Ehlers-Danlos syndrome, hypermobility type, constituting a phenotypic continuum with or, perhaps, corresponding to the joint hypermobility syndrome (JHS/EDS-HT), is likely the most common, though the least recognized, heritable connective tissue disorder. Known for decades as a hereditary condition with predominant rheumatologic manifestations, it is now emerging as a multisystemic disorder with widespread manifestations. Nevertheless, the practitioners' awareness of this condition is generally poor and most patients await years or, perhaps, decades before reaching the correct diagnosis. Among the various sites of disease manifestations, skin and mucosae represent a neglected organ where the dermatologist can easily spot diagnostic clues, which consistently integrate joint hypermobility and other orthopedic/neurologic manifestations at physical examination. In this paper, actual knowledge on JHS/EDS-HT is summarized in various sections. Particular attention has been posed on overlooked