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# Natural history and manifestations of the hypermobility type Ehlers-Danlos syndrome: a pilot study on 21 patients.

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Hypermobility type Ehlers-Danlos syndrome (HT-EDS) is a relatively frequent, although commonly misdiagnosed variant of Ehlers-Danlos syndrome, mainly characterized by marked joint instability and mild cutaneous involvement. Chronic pain, asthenia, and gastrointestinal and pelvic dysfunction are characteristic additional manifestations. We report on 21 HT-EDS patients selected from a group of 40 subjects with suspected mild hereditary connective tissue disorder. General, mucocutaneous, musculoskeletal, cardiovascular, neurologic, gastrointestinal, urogynecological, and ear-nose-throat abnormalities are investigated systematically and tabulated. Six distinct clinical presentations of HT-EDS are outlined, whose tabulation is a mnemonic for the practicing clinical geneticist in an attempt to diagnose this condition accurately. With detailed clinical records and phenotype comparison among patients of different ages, the natural history of the disorder is defined. Three phases (namely, hypermobility, pain, and stiffness) are delineated based on distinguishing manifestations. A constellation of additional, apparently uncommon abnormalities is also identified, including dolichocolon, dysphonia, and Arnold-Chiari type I malformation. Their further investigation may contribute to an understanding of the pathogenesis of the protean manifestations of HT-EDS, and a more effective approach to the evaluation and management of affected individuals.


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