Phenotypic evaluation and natural history of Ehlers Danlos Syndrome Hypermobility Type (EDS-HT)

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Ehlers Danlos Syndrome-hypermobility type (EDS-HT) (OMIM # 130020) is a connective tissue disorder characterized by hypermobility of the joints, soft skin and easy bruising in the absence of other skeletal and organ anomalies. Recently additional symptoms have been reported to have an association with EDS-HT phenotype, including gastrointestinal dysfunction, cardiac abnormalities and dysautonomia. Identification of the full phenotypic range of EDS-HT will provide insight into the disorder, allow for the creation of more effective diagnostic criteria, and ultimately better patient identification and management. In this study, we identified 103 adult patients with a clinical diagnosis of EDS-HT. The medical records of these patients were analyzed and recorded with a focus on physical characteristics, medical procedures and provider notes. From these data, statistical analyses were employed to determine significant co-morbidities and associations. Based on our findings, we described EDS-HT as having a broad clinical spectrum based on the evaluation of a large adult patient population. We report multi system involvement, as previously reported in the literature, and describe a more severely affected subpopulation characterized by dysautonomia and medical disability. These results were used to further develop the clinical phenotype and delineate the natural history of EDS-HT. Further investigation into the pathogenesis of these clinical findings is needed to help establish an effective diagnostic criterion.