

The Spine and the Craniocervical Junction in Ehlers-Danlos Syndrome

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ABSTRACT

The Ehlers-Danlos syndromes (EDS) are a heterogeneous group of hereditary disorders of connective tissue characterized by joint, skin and vascular abnormalities. Joint laxity and dislocations are commonly recognized musculoskeletal features of EDS. However, the frequency of spinal involvement, including degenerative disc disease and dural ectasia, is unknown. Previous anecdotal reports have described spinal abnormalities including spondylolisthesis, lumbar platyspondyly and scoliosis. We assessed images of the lumbar spine by Magnetic Resonance Imaging (MRI) in 58 consecutive patients with a diagnosis of EDS. The cohort included patients with hypermobile, classical and vascular forms of EDS, and the age range was 12-67. The abnormalities observed included degenerative disc disease, disc herniation, facet arthrosis, dural ectasia and dural cysts. The abnormalities were observed in all age groups, including patients as young as 15 years of age. All patients over the age of 45 had significant disc disease. 45/58 patients were found to have some degree of disc disease. 15/58 patients had dural ectasia. Degenerative disc disease was highly correlated with the presence of back pain. During the course of the clinical evaluation, it was noted that twelve patients in the cohort had a previous diagnosis of Arnold Chiari I malformation, or received the diagnosis during longitudinal follow-up. Two patients had a history of previous decompression surgery; four others were operated after enrollment in the study. The remaining patients were treated conservatively. Several additional patients had evidence of a retroflexed odontoid or pannus formation around the odontoid that was felt to contribute to complaints of head and neck pain. The results indicate that pathology in the spine and the craniocervical junction is a frequent cause of morbidity in EDS. The data suggest that it is advisable to pursue appropriate radiological evaluations in the presence of back, head and neck pain in this group of patients.