Clinical study of hereditary disorders of connective tissues in a chilean population: Joint hypermobility syndrome and vascular Ehlers-Danlos syndrome

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Objective. To demonstrate the high frequency and lack of diagnosis of joint hypermobility syndrome (JHS) and the seriousness of vascular Ehlers-Banlos syndrome (VEDS). Methods. Two hundred forty-nine Chilean patients with hereditary disorders of the connective tissues (CTDs) and 64 control subjects were evaluated for the diagnoses of JHS and VEDS using the validated Brighton criteria, as compared with the traditional Beighton score. In addition, the presence of blue sclera was determined, with the degree of intensity graded as mild, moderate, or marked. Results. The frequency of hereditary CTDs was 35%, with diagnoses of JHS in 92.4% of subjects, VEDS in 7.2%, and osteogenesis imperfecta in 0.4%. The Beighton score proved to be insufficient for the diagnosis of JHS (35% of subjects had a negative score), whereas the Brighton criteria yielded positive findings (a diagnosis of JHS) in 39% of control subjects. Blue sclera was frequent, being identified in 97% of JHS patients and 94% of VE