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The Core Curriculum

Ehlers-Danlos syndrome

Objectives

1. Describe the major clinical features of Ehlers-Danlos syndrome
2. Discuss the orthopaedic problems associated with Ehlers-Danlos syndrome
3. Discuss the genetics of Ehlers-Danlos syndrome
4. Discuss the factors underlying a high complication rate for bone and joint surgery in patients with Ehlers-Danlos syndrome

Discussion point

1. Might patients with undiagnosed Ehlers-Danlos syndrome initially present to an orthopaedic surgeon? With what types of problems?

Discussion

Ehlers-Danlos syndromes (note it is no longer officially a singular, but multiple syndromes) are a heterogeneous group of heritable connective tissue disorders characterized by joint hypermobility, skin extensibility, and tissue fragility. As clinical experience with Ehlers-Danlos syndrome accumulated, it was long evident that there was a great deal of variability in severity and expression of these disorders. Thus, a nosology was developed in which newly discovered types of the disorder would be categorized by a roman numeral, and 11 types were officially designated. Type I was the gravis type, Type II hypermobility, Type IV arterial-ecchymotic, Type VI ocular-scoliotic, etc. Joint hypermobility as an isolated disorder was often diagnosed as Ehlers-Danlos syndrome since the diagnosis was primarily based on phenotypic expression. With advances in molecular biology, a number of authorities proposed a new system of classification (Beighton,1998) to better classify data and determine the natural history of the differing syndromes under the heading of Ehlers-Danlos. The new classification includes the following types:

- Classical - autosomal dominant, skin hyperextensibility, widened atrophic scars, joint hypermobility. No consistent identifiable disorder in collagen, but deficient Type V collagen has been found in a number of families
- Hypermobility - autosomal dominant, skin hyperextensibility, recurring joint dislocations, chronic joint/limb pain
- Vascular - autosomal dominant, thin, translucent skin, arterial/intestinal/uterine fragility or rupture, extensive bruising, characteristic facial appearance. Structural defect in type II collagen encoded by COL3A1.

- Kyphoscoliosis - autosomal recessive, generalized joint laxity, severe muscle hypotonia at birth, pregressive scoliosis, scleral fragility. Deficiency of lysyl hydroxylase, and recommended laboratory test is total urinary hydroxylysyl pyridinoline
- Arthrochalasia - autosomal dominant, severe generalized hypermobility, hip dislocation. Deficient processing of Type A or Type B chains of collagen I, because of skipping of exon 6 in either gene. This may be determined by electrophoresis of dermal collagen.
- Dermatosparaxis - autosomal recessive, severe skin fragility, sagging redundant skin. Deficiency of procollagen I N-terminal peptidase, electrophoresis can quantitate.

The knowledge base of Ehlers-Danlos syndromes is obviously in a developmental stage, and further genetic and molecular analysis will clarify some of the confusion resulting from the inability to accurately classify these syndromes. Obviously, depending on the type, vascular, skin healing, recurrent dislocation, and other complications will be more frequent when performing surgery on this group of patients.

References

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