



POSNA

The Core Curriculum

Marfan syndrome

Objectives

1. Describe the genetic basis of Marfan syndrome
2. List the skeletal diagnostic criteria for diagnosis of Marfan syndrome from the Berlin 1986 nosology
3. Discuss major factors affecting mortality of patients with Marfan syndrome
4. Discuss the orthopaedic problems most often encountered by patients with Marfan syndrome

Discussion points

1. Why is it sometimes difficult to make the diagnosis of Marfan syndrome?
2. What problems have been reported in the spine of patients with Marfan's syndrome?

Discussion

Marfan syndrome is presently undergoing intense investigation, especially from the genetic and molecular perspective. It is now accepted that mutations in the gene for fibrillin-1 (FBN-1) have been shown to cause Marfan syndrome, an autosomal dominant disorder of connective tissue. Fibrillin-1 is a component of the microfibrils which accompany elastin in the elastic fiber. The exact function of the microfibrils is currently under investigation. What is known is the phenotypic expression of the dysfunctional microfibrils, manifested by disorders of the cardiovascular system, musculoskeletal system, and ectopia lentis. Ultrastructural changes in elastic fibers have been illustrated by Gigante. Life expectancy has risen dramatically in the past generation, largely as a result of advances in vascular surgery. Dilatation of the aortic root is responsible for most cases of aortic incompetence and replacement of the root has become very successful. Neonatal Marfan syndrome is more lethal than the later onset variety.

The diagnosis of Marfan syndrome can be complex. Many of the diagnostic criteria are age dependent. There are 8 specific musculoskeletal criteria: chest wall deformity, vertebral column deformity, arachnodactyly, high arch palate, tall stature, limb disproportion, abnormal joint mobility, and protrusio acetabuli. Four (4) of these have been suggested to qualify the musculoskeletal system as a major criterion. Tall stature is most frequent. A clinical test for arachnodactyly is the Steinberg test, where the thumb protrudes past the ulnar border of the palm when opposed in the fist. There are a number of "fibrillinopathies" which have variable expressions of the major diagnostic features of microfibril dysfunction. In addition, congenital contractural arachnodactyly has been identified as a type 2 fibrillinopathy.

Joint hypermobility and spine hypermobility, with a higher than normal rate of atlantoaxial subluxation, spondylolisthesis, kyphosis, and scoliosis are characteristic of Marfan syndrome. The atlantoaxial subluxation is less dramatic than that associated with Down's syndrome. Dural ectasia is common in Marfan syndrome and may be associated with back pain.

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