Musculoskeletal case 15. Diagnosis

Marfan's syndrome

The diagnosis was Marfan's syndrome complicated by an extensive aortic dissection extending distally to involve the iliac vessels. The association of a tall, slender build with spinal deformity, aortic dissection and dural ectasia should suggest the diagnosis. Plain radiographs of the hands showed evidence of arachnodactyly with a positive metacarpal index of 10, helping to confirm the diagnosis. (Fig. 5 is a radiograph of the hand in patient with classical Marfan's syndrome. The fingers are long and slim.) The metacarpal index is measured by adding the lengths of the metacarpal bones and dividing by their diameter. A positive metacarpal index, as in this case, is said to be present when the index is greater than 8.4.

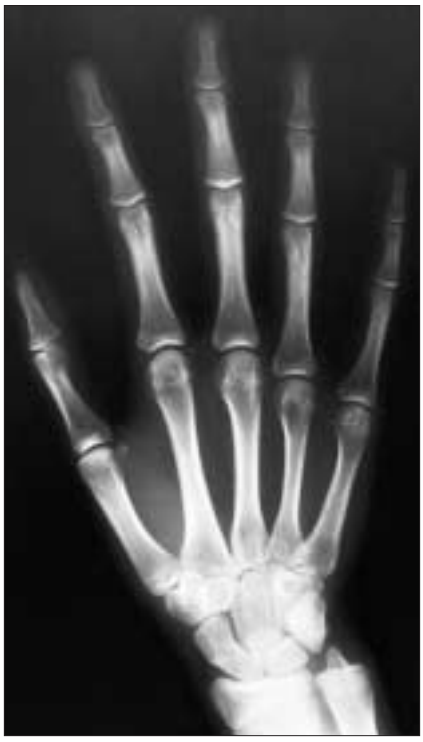

FIG. 5.

Marfan's syndrome is an inherited disorder of connective tissue that affects many organ systems, including the skeleton, lungs, eyes, heart and blood vessels.[1] It is an autosomal dominant disease with a single abnormal defect of the gene *FBN1* that encodes fibrillin-1 and determines the structure of fibrillin, a protein that is an important component of connective tissue.[2] Approximately one-quarter of the cases occur as a result of a spontaneous mutation. The syndrome was first described approximately 100 years ago and remains predominantly a clinical diagnosis. The condition affects men and women of all races and ethnic groups. People with Marfan's syndrome are typically very tall, slender and loose jointed. Their arms, legs, fingers and toes may be disproportionately long in relation to the rest of the body. They often have a long, narrow face, and the roof of the mouth may be arched, causing the teeth to be crowded. Other skeletal abnormalities include pectus excavatum or carinatum, spinal scoliosis and pes planus.[3]

More than half of all people with Marfan's syndrome experience dislocation of one or both lenses of the eye. Many are also myopic, and there is an increased association with glaucoma and cataracts. Dural ectasia is common and in some cases may lead to a spine-related neuropathy. Within the lungs pneumothoraces are more common as well as bullous lung disease and emphysema.

The most serious problems associated with Marfan's syndrome involve the cardiovascular system. Aortic and mitral valve involvement is common, leading to incompetence. Aortic dissection is a potentially life-threatening complication that may require surgery, depending on the site of involvement.[4] Pregant women with Marfan's syndrome run a 1% risk of a fatal complication,[5] and this risk rises with increasing aortic root diameter. Cesarean section at 38 weeks' gestation should be offered if aortic root diameter is more than 4.5 cm. Over the past 30 years, evolution of aggressive medical and surgical management of the cardiovascular problems, especially mitral valve prolapse, aortic dilatation and aortic dissection, has resulted in considerable improvement in life expectancy.

References

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