



POSNA

The Core Curriculum

Fibrous dysplasia

Objectives

1. Describe differing patterns of involvement noted in patients having fibrous dysplasia of bone
2. Describe the pathology of fibrous dysplasia
3. Discuss the genetic anomalies associated with fibrous dysplasia
4. Discuss the natural history and results of treatment of bony deformity secondary to fibrous dysplasia

Discussion points

1. What bones are most often affected with fibrous dysplasia?
2. Why is fibrous dysplasia so difficult to treat effectively?

Discussion

Fibrous dysplasia is an annoying and intriguing benign bone lesion. It is not a neoplasia, but a developmental abnormality. It can present in a monostotic or polyostotic form. The polyostotic form generally is more severe and tends to be unilateral, the monostotic is fortunately more common. Albright's syndrome consists of cutaneous pigmentation, polyostotic fibrous dysplasia, and precocious puberty in girls. Other endocrine abnormalities have been noted with the polyostotic form. Fibrous dysplasia overall has a slight female preponderance. Radiographically, the appearance is somewhat variable. The lesion is usually diaphyseal. The cortex is expanded, with the "ground glass" appearance of the medullary canal. Calcification may be present. Radionuclide scans are markedly positive, out of proportion to the radiographic appearance. Microscopically, fibrous dysplasia has a very characteristic appearance, with irregular strands of osteoid and bone in a background of fibrous tissue. The lesions of fibrous dysplasia are difficult to treat. Curettage is ineffective, especially in childhood. If cancellous graft is placed, it will be absorbed. Enneking and Gearen recommended replacement of the lesion with cortical allograft, which would resist absorption by the host bone and provide structural stability. Guile noted that the lesion never healed, and the only way to improve function was with valgus osteotomy of the proximal femur to improve the biomechanics, essentially ignoring the lesion. Vascularized bone grafting has been reported as effective for lesions of the upper limb.

Fibrous dysplasia is of interest from a genetic standpoint, because it appears to be a somatic mutation, in that the mutation occurs after fertilization in some subsequent cell division. The site of the mutation has been located, the gene for the alpha subunit of

stimulatory guanine-nucleotide-binding protein, a protein that stimulates cyclic adenosine monophosphate is mutated. This defect has been found both in polyostotic and monostotic forms of fibrous dysplasia. The ultimate therapy for fibrous dysplasia will very likely be derived from recombinant protein resulting from further investigation of the genetic control of this condition. Pamidronate has recently been shown to have a beneficial effect on the ability to successfully instrument bony lesions of fibrous dysplasia associated with McCune-Albright syndrome. Malignancy has been documented in about 2% of cases followed at the Mayo clinic. Prior radiation was associated with some, but not all of the lesions. Radiation therapy does not appear to be a logical treatment at present.

References

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