



POSNA

The Core Curriculum

Hereditary multiple exostoses (HME) (Diaphyseal aclasia)

Objectives

1. Describe the clinical features of HME
2. Describe the genetic transmission of HME
3. Discuss the predisposition to formation of chondrosarcoma in patients with HME
4. Discuss indications for excision of osteochondromata in patients with HME
5. Describe complications of excision of osteochondromata in patients with HME

Discussion point

1. Is HME a dysplasia or neoplasm?

Discussion

Autosomal dominantly transmitted HME, although a single phenotype, has been linked to three different chromosomal locations, which comprise a family called the EXT genes. EXT genes are felt to be tumor suppressor genes, thus mutations would render the patient more susceptible to neoplasia. Both alleles must be mutated for an exostosis to occur. HME has been classified as a skeletal dysplasia, but a recent paper proposes that HME is a familial neoplastic trait, and compares the behavior of HME to familial colorectal polyposis. It was further suggested that osteochondromata result from clonal expansion of single cells into osteochondromas that disrupt further growth, and that excision might inhibit subsequent deformity. Efforts have also been recently made to classify HME on the basis of the type and severity of forearm deformity. Greater deformity accompanied sessile lesions. The relationship of patterning of osteochondromata to potential for malignant degeneration is presently being actively investigated. The rate of malignancy in 175 patients followed for over a 40 year period was 0.57%, but other series have reported a rate up to 25%. Clearly, patterns of genetic transmission can vary between geographic sites, so determination of a single rate is probably an unreasonable goal.

HME is consistently recognized during childhood. The most frequent deformities resulting involve the wrist from ulnar shortening, ankle and knee valgus. Physeal stapling, excision of lesions, osteotomies, and lengthenings have all been performed in various combinations for deformity. An impressive list of vascular complications, predominately about the knee, of osteochondromata has been reported by Vasseur, with the admonition that prophylactic resection of lesions near a vessel should be performed. The complication rate of excising osteochondromata is 12%, mostly peroneal neuropraxia, but arterial laceration also occurred.

The indications for intervention for wrist deformity are conflicted. Peterson advocates an aggressive approach; Arms and Stanton are less inclined to intervene.

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