Dysplasia epiphysealis hemimelica presenting as a progressive, painful flatfoot deformity[1]

Michael Levine, M.D.
Alan R. Gurd, M.D.

A case of dysplasia epiphysealis hemimelica is presented involving a growing child with a progressive, painful flatfoot deformity. A characteristic of this disorder is the osteocartilaginous overgrowth of the navicular and talus in a single extremity, along with pain and deformity as presenting complaints. Symptoms were relieved with excision of the exostosis.

Index terms: Bone diseases, developmental · Case reports · Flatfoot

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The flatfoot is a common problem seen by both the pediatrician and orthopedic surgeon. Frequently, it is secondary to a flexible plantar flexed talus and is merely a postural-type problem. Flatfoot has been classified as either a static, arthritic, or neuromuscular deformity. There are times when the foot problem is the result of a bony abnormality and results in a rigid painful condition.[1]

Here we report a case of dysplasia epiphysealis hemimelica—a developmental disorder characterized by the asymmetrical overgrowth of one or more epiphyses in a limb or tarsal or carpal bone in childhood.[2] This case involved the talonavicular joint and presented as a painful, progressive flatfoot deformity

Case report

A four-year-old boy was referred to The Cleveland Clinic Foundation with a five-month history of a progressive flatfoot deformity and a tender bony prominence along the medial aspect of the right foot. During the summer of 1983, he injured this foot in a bicycle accident. Evaluation by a local physician suggested a hairline fracture in the foot, which was treated with an Ace bandage and nonweight-bearing ambulation with crutches for two weeks. The radiographs were not available for evaluation. After two weeks of conservative therapy, the pain resolved and the boy resumed his normal activities.

He did well until December 1983, when he began complaining of pain along the medial aspect of the right foot. His parents additionally noted that the foot was becoming flat and there was a prominence along the medial aspect. He was evaluated and radiographs were obtained by a local physician who was concerned about the possibility of a tumor. His family history was negative for developmental disorders, and his past medical history was negative. His general physical examination was essentially unremarkable. Examination of his right foot and ankle revealed a rigid pes planus deformity with a palpable tender mass in the medial arch. Heel valgus with abduction of the forefoot was also noted. The peroneus longus tendon was intact and not spastic. His neurological examination was unremarkable, as were his laboratory-test results at the time of admission. Radiographs showed normal bony structures of his right ankle with a normal ankle joint mortis. Views of the right foot demonstrated deformities in the medial aspect of the talus as well as the tarsal navicular bone. The medial aspect of these structures had a lobulated appearance with several protruding bony ossicles (*Fig. 1*).

On April 4, 1984, surgical exploration of the right foot was performed with excision of multiple exostoses of the right talonavicular joint. A synovial biopsy was done and a short leg cast was applied. The posterior tibial tendon was

[1] Department of Orthopaedic Surgery, The Cleveland Clinic Foundation. Submitted for publication Mar 1986; accepted June 1986.

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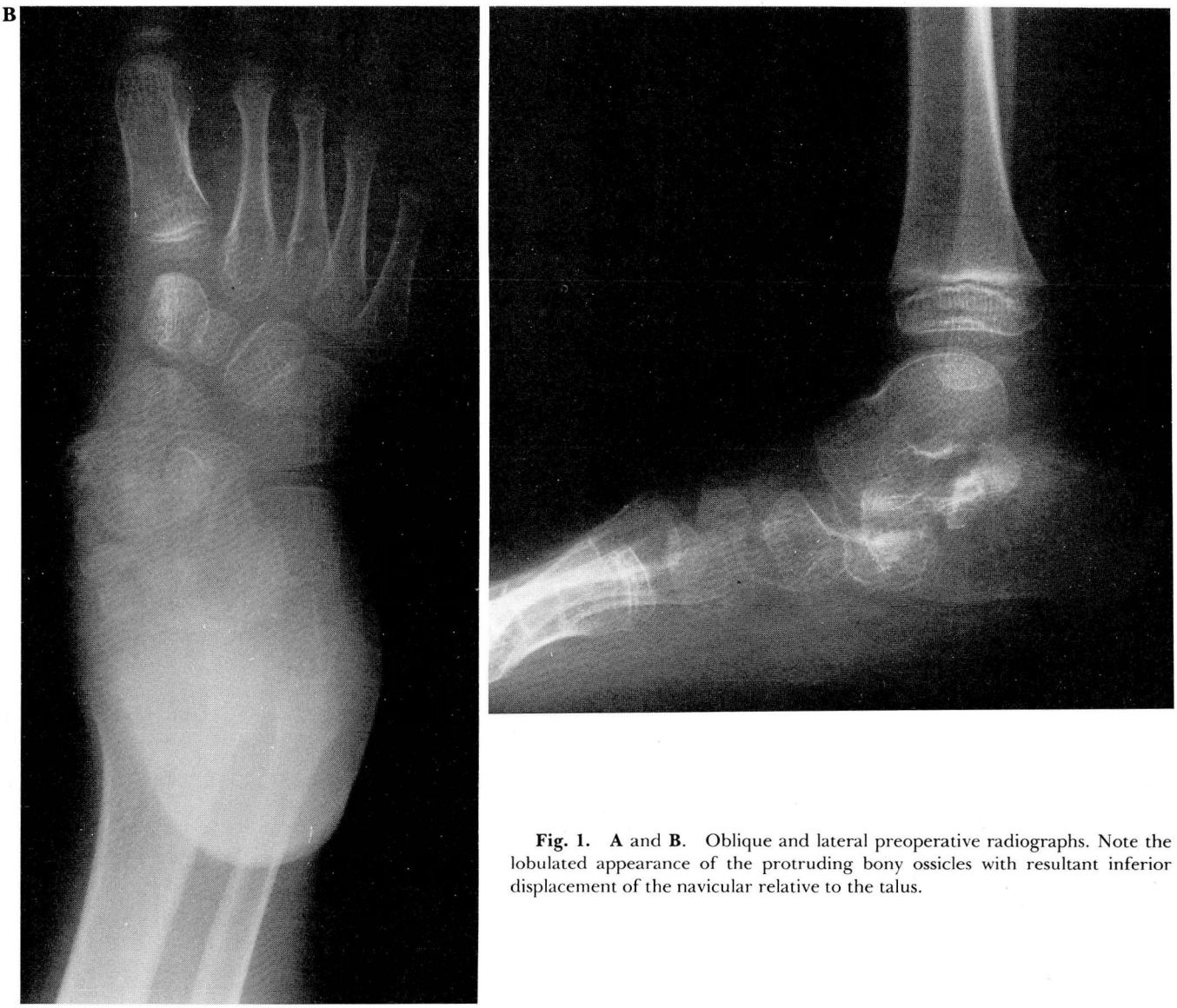

Fig. 1. **A** and **B**. Oblique and lateral preoperative radiographs. Note the lobulated appearance of the protruding bony ossicles with resultant inferior displacement of the navicular relative to the talus.

intact and not reefed. Intraoperative findings revealed multiple fragments of normal-appearing bone with cartilaginous caps attached primarily to the talus and navicular, but also the calcaneus, resulting in a lateral displacement of the talus on the navicular (*Fig. 2*).

Pathologic findings revealed a reactive synovial proliferation. The bone was histologically normal. The pathologic diagnosis was consistent with multiple osteochondromas or dysplasia epiphysealis hemimelica. A diagnosis of dysplasia epiphysealis hemimelica was made based on the typical radiographic and pathologic findings.

His postoperative course was unremarkable. On his return visit two weeks following surgery, the radiographs showed significant improvement with no apparent abnormal fragments. He was initially given a UCBL orthosis to maintain his reformed arch during weight-bearing activities. Upon follow-up 15 months later, the patient was asymptomatic, with a mobile subtalar joint and no recurrence as shown radiographically (*Fig. 3*).

Discussion

The first case of epiphysealis dysplasia hemimelica was referred to as "tarsomegalie" by Mouchet and Belot in 1926.[3] In 1950, Trevor[4] further described the disorder and called it "tarso-epiphyseal aclasis." The present term was given by Fairbank in 1956.[5]

The etiology of dysplasia epiphysealis hemimelica remains unknown. Hensinger et al[6] described a family in which it appeared to be inher-

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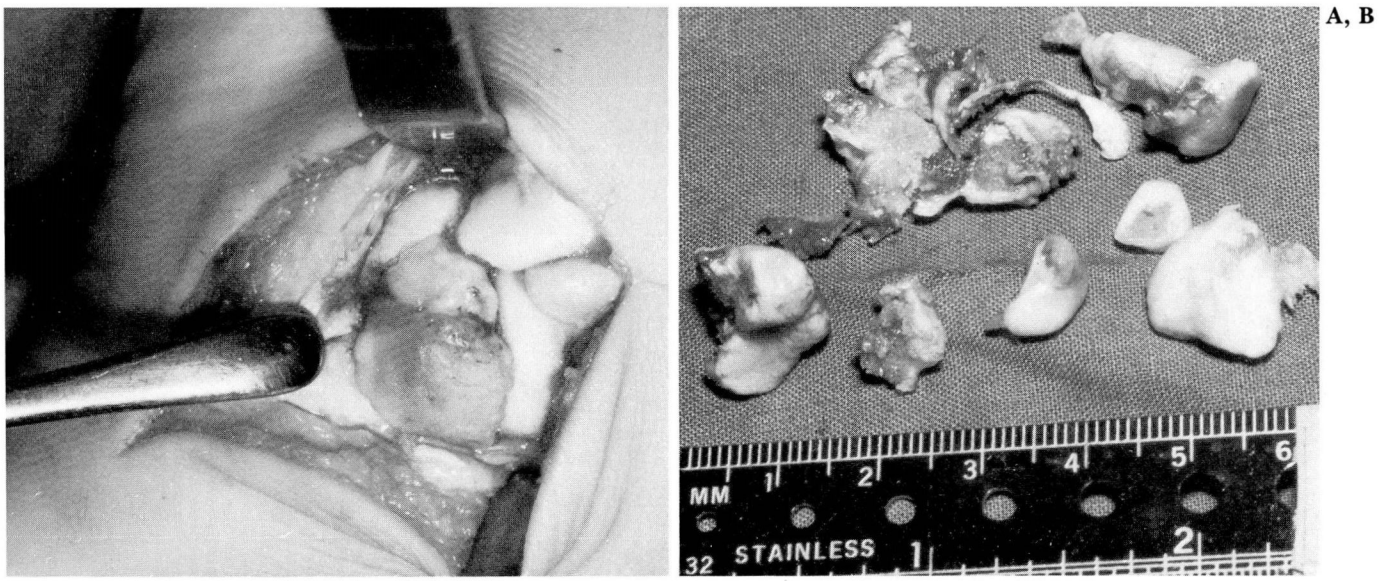

Fig. 2. A. Intraoperative photograph showing multiple bony ossicles within the area of the talonavicular joint. **B.** Pathologic specimen.

ited as an irregular autosomal dominant trait. But Connor et al,[2] in a retrospective series with long-term follow-up, were unable to document a genetic etiology or any common environmental factor.

Dysplasia epiphysealis hemimelica is primarily a developmental disorder with most cases presenting between the ages of two and 14, but with reported extremes from birth to age 63.[7] Males are affected three times more often than females.[8]

The usual presenting features of the disorder are painless swelling or deformity in an otherwise healthy individual.[5,8] Kettlekamp et al[8] noted that pain was a common presentation after a fracture of an exostosis. Additionally, Fairbank[5] noted that the mother may often complain that the child has a progressive unilateral flat foot, which is similar to the case described here. Kettlekamp et al[8] also showed in a series of 15 cases that when the lesion involves the lower extremity, the medial side is affected twice as often as the lateral. The talus, distal femoral, and distal tibial epiphyses are the most common sites.

The radiographic appearance of epiphysealis dysplasia hemimelica has been described as an irregular, lobulated, osseous mass extending from the epiphysis, tarsal or carpal bone—similar to an exostosis.[8] With tarsal involvement, the talus, as well as the navicular and cuneiform bones, may be markedly enlarged, although this was not really appreciated in our case. However, involvement of the calcaneus, which *was* noted in this case, is uncommon and believed to be secondary to the talar abnormality.[5]

Pathologically, the appearance is similar to an osteochondroma with no characteristic feature to differentiate between the two. The diagnosis, therefore, is based on the age of the patient, the regions involved, and the radiographic appearance.[5]

The treatment to date has been that of surgical excision of the lesions causing deformity or pain. Kettlekamp et al[8] reported improvement for the 12 patients in their series who underwent surgical excision. Since we noted local recurrence in only 2 of 15 patients, complete removal of the exostoses seems essential. Close follow-up of these patients is required, for the potential of cartilage proliferation exists well into adult life.

Review of this case reveals consistencies with those previously described in the literature. Following surgical exicision of the exostoses, the painful flatfoot deformity markedly improved. The exostoses has not recurred to date.

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A, B

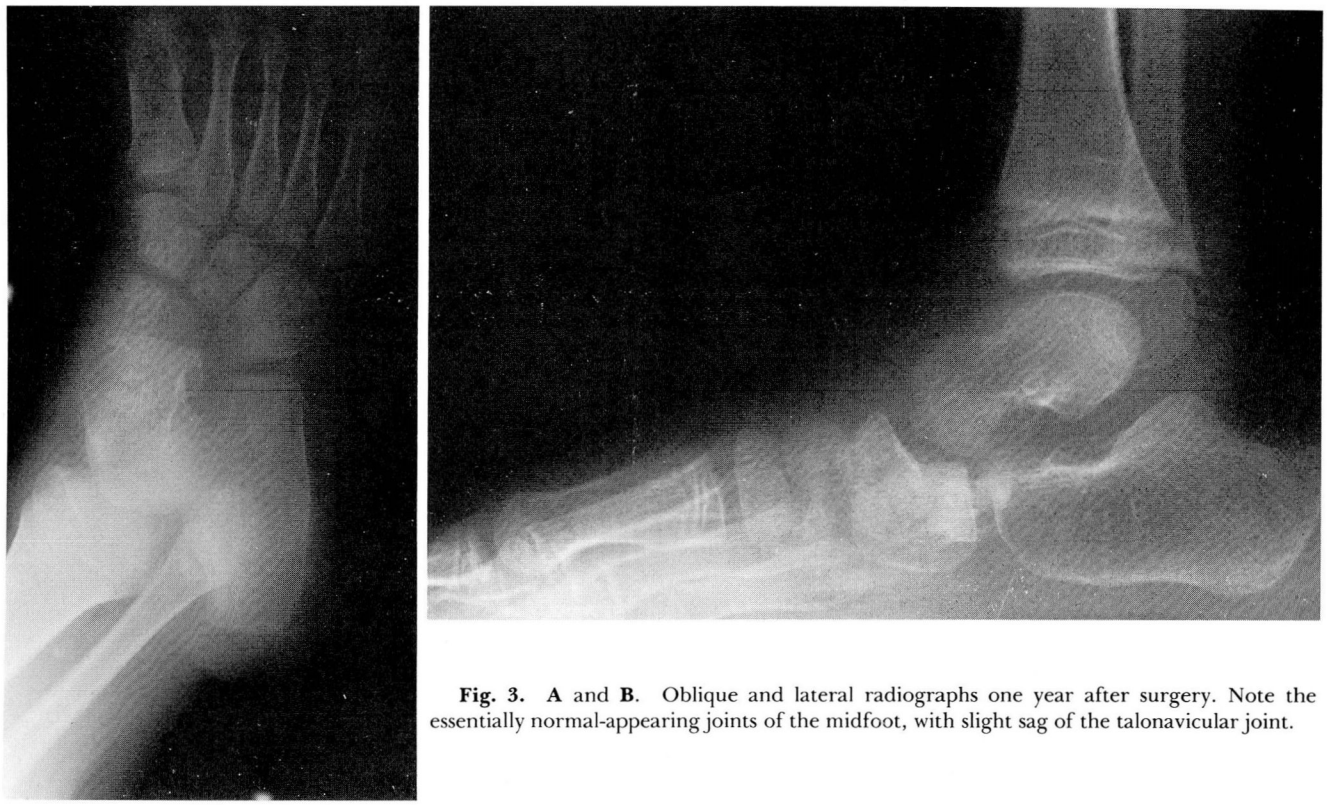

Fig. 3. **A** and **B**. Oblique and lateral radiographs one year after surgery. Note the essentially normal-appearing joints of the midfoot, with slight sag of the talonavicular joint.

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Alan R. Gurd, M.D.
Department of Orthopaedic Surgery
The Cleveland Clinic Foundation
9500 Euclid Ave.
Cleveland, OH 44106

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