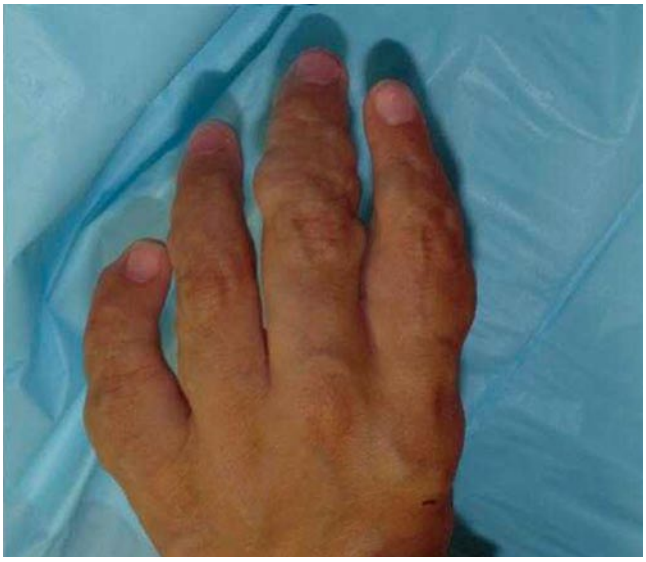
What Is Your Diagnosis?



A 5-year-old girl being followed by the orthopedic service for a pathologic fracture of her left femur presented with new 3- to 5-mm compressible purple nodules on the lateral aspects of the digits on her left hand.

PLEASE TURN TO PAGE 115 FOR DISCUSSION

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The Diagnosis: Maffucci Syndrome

Maffucci syndrome is a rare genodermatosis characterized by multiple enchondromas (Figures 1 and 2A) and venous malformation of the skin and mucous membranes (Figure 2B). This syndrome was first described by Maffucci[1] in 1881. It affects males and females equally and does not appear to have any racial predilection.[2,3] Maffucci syndrome does not demonstrate any simple mendelian genetic pattern of inheritance. The average age of onset is 4 years (range, birth to 30 years). In approximately 27% of cases, the patient has bony or soft tissue abnormalities at birth.[4] Intelligence is unaffected and karyotypes usually are unaffected.[5-10] Fertility is not affected and females with the syndrome have delivered unaffected children.[11-13]

The enchondromas are thought to be part of a generalized mesodermal dysplasia. As bones grow and lengthen, cartilage is left behind and grows irregularly, which results in enchondromas. Deformities, shortening of extremities, and pathologic fractures occur in approximately 20% to 26% of cases.[2,4] The enchondromas are found in all parts of the skeleton including the hands (87%–89% of cases), feet (36%–61% of cases), tibia/fibula (52%–59% of cases), femur (36%–54% of cases), humerus (34%–43% of cases), radius/ulna (29%–42% of cases), ribs (27%–32% of cases), pelvis (21%–25% of cases), scapula (20%–26% of cases), and head (8%–18% of cases).[3,4]

The hemangiomas present as blue subcutaneous nodules that can be emptied by pressure or by elevating the lesions above the heart.[2] They also can be found throughout the body including the hands (57% of cases), feet (41% of cases), arms (39% of cases), legs (38% of cases), trunk (29% of cases), and head/neck (25% of cases).[4] In rare cases, they also have been found in the leptomeninges,[5] eyes,[14] pharynx,[9,15,16] tongue,[17] trachea,[18] and intestines.[19] Thrombi often form within the hemangioma, causing calcified lesions called *phleboliths*. The hemangiomas most often are venous in nature, but capillary and mixed types can occur.[2,20]

Malignant transformation occurs in approximately 23% to 37% of cases.[3,4] The most common malignant transformation is enchondromas changing into chondrosarcomas, which is believed to occur in 15% to 30% of cases.[3,4,21] Other associated malignancies include fibrosarcomas[22]; angiosarcomas[3,23,24]; lymphangiosarcomas[25,26]; osteosarcomas[27]; mesenchymal ovarian tumors[3]; gliomas[3]; and breast,[25] pancreatic,[22] and liver adenocarcinomas.[28]

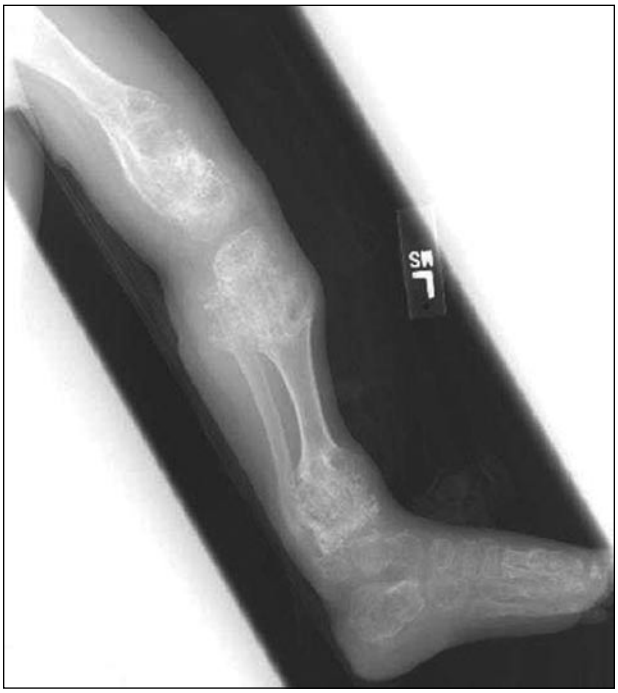

Figure 1. Radiograph of left leg showing a pathologic fracture of the femur.

Hemangiosarcomas are extremely aggressive tumors that are often fatal. They frequently recur, metastasize early, and have a median survival time of 20 months.[29] Lymphangiosarcomas also are very aggressive with a mean survival time of less than 31 months and a 5-year survival time in less than 15% of cases.[30,31] Once any of the above neoplasms develop in a patient with Maffucci syndrome, the risk for developing a second neoplasm increases.[4]

Mildly affected patients with Maffucci syndrome do not require treatment. Moderately affected patients may need to wear special shoes or use crutches because of bone deformities. Severely affected individuals may even require functional amputation at the transfemoral or transhumeral head and the use of prosthetic devices.[32] Radiation therapy has been employed in the past to reduce the size of hemangiomas, but the benefits are questionable and the modality is not recommended.[3] Because of the possibility of hemangiomas undergoing malignant transformation, it is important to perform biopsies of lesions that enlarge rapidly, continue to grow after the patient is full grown, or become painful. Excisional biopsy is preferred because the entire lesion is available for examination and hemostasis is easier to obtain.[3]

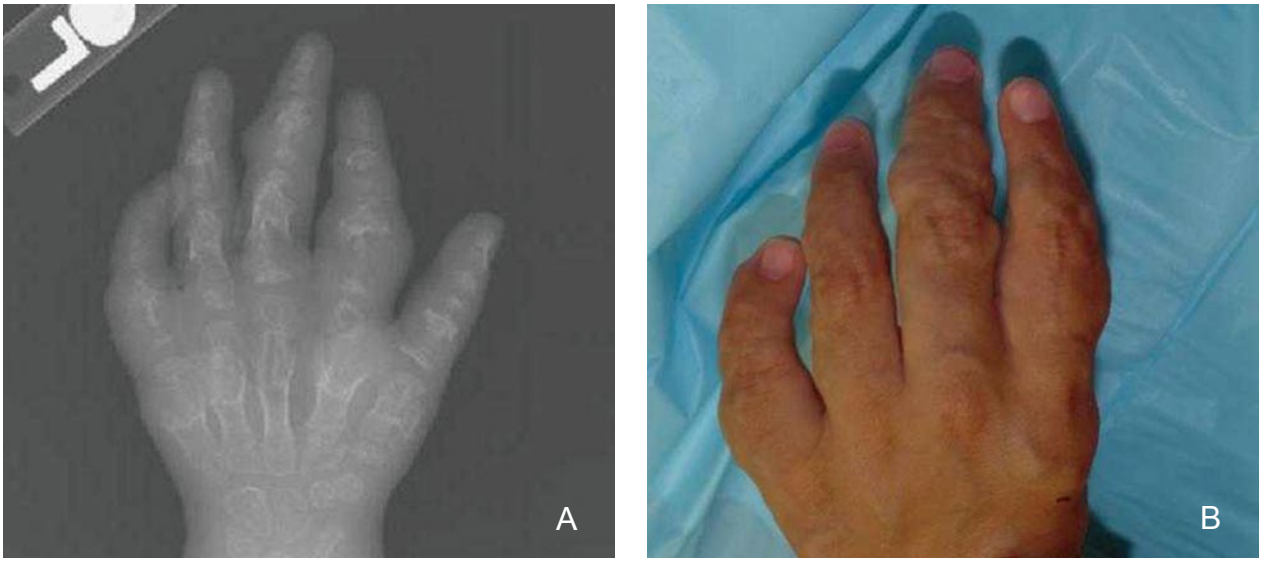

Figure 2. Radiograph of left hand demonstrating multiple enchondromas (A). Left hand with multiple compressible purple nodules demonstrating venous malformations (B).

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