Tuberous sclerosis of the tibia

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Section: Musculoskeletal system
Case Type: Clinical Cases
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Patient: 12 years, female

Clinical History:

A 12 year old girl with known tuberous sclerosis presented with a valgus deformity of the left tibia.

Imaging Findings:

A 12 year old girl with known tuberous sclerosis presented with a few months history of a worsening valgus deformity of the left lower leg. She had had previous neurosurgical procedures including removal of a giant cell astrocytoma from the brain, and insertion of a VP shunt to relieve bilateral hydrocephalus. She had cognitive impairment and was on medication for epileptic seizures. Plain radiographs were performed of the affected limb. They confirmed the valgus deformity and showed periosteal thickening of the left tibia. The left fibula was also affected to a lesser extent.

Discussion:

Tuberous sclerosis is an autosomal dominant condition classically characterized by a clinical triad of epileptic seizures, mental retardation and adenoma sebaceum. This triad is found in only 30% of patients and the disorder has a wide spectrum of phenotypic expressions with hamartomatous malformations occurring mainly in the brain, kidney, lung, skin and heart. The extracranial skeleton may be focally or diffusely involved with radiolucent cyst-like lesions or dense sclerotic deposits. Irregular subperiosteal new bone formation can produce a thickened undulating cortical contour. Cortical involvement is usually seen in the short tubular bones of the hands and feet, and occasionally, the long tubular bones. Radiolucent cyst-like lesions mainly occur in the distal phalanges of the hand. Significant deformity is treated with orthopaedic procedures, and a tibial osteotomy will be performed on this individual. The prognosis for restoration of skeletal function is good. The differential diagnosis of the plain radiographs is tuberous sclerosis or neurofibromatosis. Previous infection or trauma is less likely.

Differential Diagnosis List: Tuberous sclerosis of the tibia (and fibula).

Final Diagnosis: Tuberous sclerosis of the tibia (and fibula).

References:

Description: Valgus deformity of the left tibia with diffuse periosteal thickening. The fibula is affected to a lesser extent. Origin:
Description: Lateral view confirms the periosteal thickening of the left tibia. Origin: