History
2 month old female with bowed right leg.

Diagnosis
Tibial Dysplasia-Neurofibromatosis Type I

Discussion
Neurofibromatosis type I is a disorder related to a single large gene on chromosome 17. The gene product, neurofibromin, includes a domain with GTPase-Activating Protein function which regulates the intracellular signal protein Ras. Then NF-1 gene is probably a tumor suppressor although loss of one allele probably accounts for many of the non-tumor manifestations including osseous dysplasia. NF-1 is a highly variable disorder with manifestations often beginning at birth. Phenotypic features can be divided into tumor (eg, nerve sheath tumors, glial series neoplasms, leukemia, pheochromocytoma, and rhabdomyosarcoma) and non-tumor (eg, cafe-au-lait spots, cognitive disability, vascular dysplasia, bone dysplasia). Thin gracile ribs, sphenoid wing dysplasia, congenital bowings and pseudoarthrosis, and scoliosis can be seen in NF-1. Osteoblasts, osteoclasts, chondrocytes, fibroblasts and endothelial cells express neurofibromin and may be functionally compromised when neurofibromin is deficient.

Anterolateral bowing of the tibia with or without a hypoplastic fibula, focal narrowing and intramedullary sclerosis or cystic change at the apex of the bowing are characteristic of neurofibromatosis type I. Mesodermal dysplasia produces deficient osteofibrous hamartomatous tissue which is susceptible to pathologic fracture and possibly nonunion and pseudoarthrosis. Prophylactic bracing may be used to prevent development of fracture; if fracture occurs, osteotomy with bone grafting may be necessary.

Findings
CR-Marked anterolateral bowing of the mid-tibial diaphysis irregular cortical thickening and distortion of the medullary bone. The fibula is thin undulating.

Reference
Schindeler A, Little DG. Recent insights in bone development, homeostasis and repair in type 1 neurofibromatosis (NF-1). Bone (2008); 42:616-622.
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