Case report 765

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Imaging studies

Figs. 1–4. See next page for corresponding captions.
Clinical information

An 18-year-old woman with an established diagnosis was admitted for possible liver transplantation. The diagnosis of type I glycogen storage disease was established at 1 week of age by liver biopsy, done for the evaluation of hypoglycemia and seizures. At age 17 years, the patient began having episodes of nonbloody, nonbilious vomiting several times a week. Abdominal sonography and subsequent computerized tomography demonstrated a 13 x 10 x 9 cm mass in the anterior segment of the right lobe of the liver (Fig. 1). A needle biopsy of the mass was consistent with adenoma. Subsequent sonograms demonstrated enlargement and heterogeneity of the mass, raising concerns about malignancy and prompting admission to the hospital.

During this admission, a radiograph of the left hand and wrist was obtained for bone age and was assessed as delayed (14 years), based upon the standards of Greulich and Pyle [1]. Deformity of the distal end of the ulna was noticed, and a skeletal survey was obtained. The distal aspects of both ulnae were foreshortened with medial bowing of the ulnae ("pseudomadelung" deformity; Fig. 2). Mild notching was present at the medial aspects of the proximal humeri. The spine had a mild to moderate thoracolumbar scoliosis with a compensatory thoracic curve. The vertebral bodies were tall and anteriorly scalloped, and the lumbo-sacral lordosis was exaggerated (Fig. 3). Bilateral coxa valga was present (Fig. 4). In general, the long bones were gracile, but generalized osteopenia was not evident. Growth arrest lines were present in the distal aspects of the tibiae.