Fucosidosis

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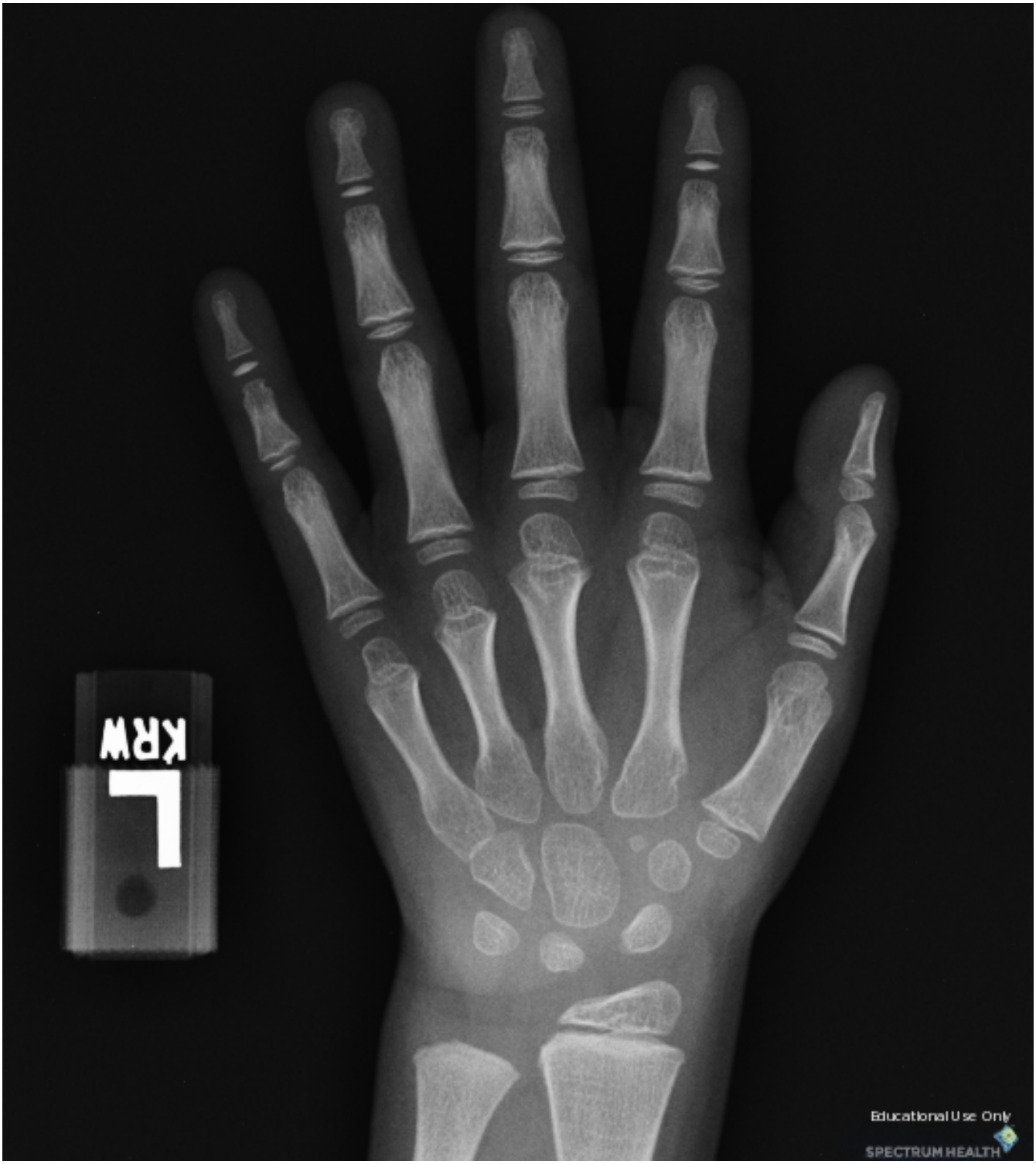
History
9 year old male with short stature and history of bone marrow transplantation.

Diagnosis
Fucosidosis

Discussion
Fucosidosis is a rare autosomal recessive disorder related to a defect on chromosome 1 leading to accumulation glycolipids, lipoproteins, oligosaccharides and polysaccharides in lysomes of various organs.
Clinical manifestations include coarse facies, prominent forehead, hypertelorism, large tongue, thick lips, heavy eyebrows, dental anomalies, broad chest, accentuated lumbar lordosis, hepatosplenomegaly, deteriorating psychomotor milestones, peripheral neuropathy, weakness, thick and dry skin, and recurrent respiratory infections. Fucose rich granules and inclusion granules are seen on histochemical evaluation.
Radiographic findings are similar to mucopolysaccharidosis. The skull is thickened with poorly developed sinuses. Platyspondyly, short odontoid process, thoracolumbar kyphosis and anterior beaking of the thoracic and lumbar vertebral bodies can be seen. The ribs may be broad and the clavicles may be shortened. In the pelvis, the acetabula may be flat and sclerotic with coxa magna. The long bones can be widened with thin cortices. Skeletal maturation is usually delayed.

Findings
CR-PA view of the left hand for assessment of bone age reveals a bone age of 6 years (chronological age is 9 years 3 months). Note the sclerotic ("ivory") epiphyses of the 2nd, 4th and 5th distal phalanges and 5th middle phalanx and mild ulna minus deformity.

Reference
Taybi H and Lachman RS. Radiology of syndromes, metabolic disorders and skeletal dysplasias, 4th Ed. Mosby (1996).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