Polyostotic Fibrous Dysplasia (A Case Report)

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POLYOSTOTIC FIBROUS DYSPLASIA
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JOHN R. CALDWELL, M.D.*

In 1937, Albright et al., described a syndrome "Osteitis Fibrosa Disseminata with Areas of Cutaneous Pigmentation and Endocrine Dysfunction with Precocious Puberty in Females." The tendency to use the name Polyostotic Fibrous Dysplasia, suggested by Lichtenstein, has come into general use for the bone disease rather than Osteitis Fibrosa Disseminata.

The case here presented shows café au lait pigmentation and multiple sites of cystic or hyperostotic bone disease but no precocious menstruation and therefore might well be designated as in the title or as Albright's Syndrome sine Precocious Puberty.

The patient, D.S., case number 483894, a girl, was first admitted**, at age 11 in August of 1946 because of a limp on walking and a waddling gait. At birth she had areas of light tan pigmentation of the skin, otherwise was alright until age 6 when she fell from a bicycle and thereafter began to limp on walking. She had not yet started to menstruate when first seen here.

On examination the pigmentation was sharply demarcated from normal skin by irregular, jagged margins and was distributed over the left anterior hemithorax with its medial margins at the midsternal line. It was also present on the left arm and thorax and lumbar regions posteriorly. There was asymmetry of the face with a bony prominence lateral to the right eye and slight proptosis of the right eye. There was prominence of the trochanters bilaterally with restriction of abduction and rotation of the hips and some atrophy of the right thigh and leg. She had some axillary hair, but no pubic hair at this time. There was slight cardiac enlargement and a grade two, apical, systolic murmur, but otherwise the physical examination was normal.

X-rays showed multiple areas of bone thickening and increased density, especially along the base of the skull, the occiput, right orbit and elsewhere in a spotty, widespread distribution. Multiple cystic areas especially involved both ilia, both femora with a very definite coxa vara deformity on both sides and other areas of thinning of the cortex in the left femur, and an incomplete fracture through the neck of the left femur. X-rays of the teeth showed normal lamina dura and there was normal bone structure between the areas of disease and generally in areas not involved.

Laboratory studies summarized for determinations on numerous occasions were as follows: (expressed in milligrams percent)—serum calcium 9.7; 10.2; 9.0; 9.6. Serum phosphorous 2.29; 2.48; 4.3; 3.37. Alkaline phosphatase 10.8; 11.0; 7.6; 31.2. A BMR was zero. A calcium balance study done over an eight-

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een day period revealed positive calcium balance. The average daily intake was 1.234 Gm., daily average fecal output was .913 Gm., and daily average urinary output was .042 Gm., giving a daily retention average of .279 Gm.

Histopathology of tissue removed from the femoral heads revealed a marrow completely replaced by cellular fibrous tissue. Bone trabeculae passed irregularly through this fibrous tissue. There were varying areas of osteoblastic and osteoclastic activity and in a few areas cartilage undergoing transformation into bone.

Three successive operations were performed for correction of the coxa vara deformity in 1947, 1948 and 1950, by the Department of Orthopedic Surgery. The final result was achieved after a procedure which involved curettment of the cystic bone lesions, replacement of the cavity with cancellous bone chips and insertion of a blade plate in the left femoral head and a Jewett nail plate with screw fixation in the right femora.

Photo of patient at age 11 showing asymmetry of face with bony prominence lateral to right orbit.

In 1951 it was evident that she had a good functional result from the corrective surgery. She could climb stairs without support other than the railing and x-rays showed some callus formation around the bone at the site of the metal plate fixation.

She returned for a progress visit in January of 1954 and reported that she had attended school regularly and would graduate in June. She was getting along well at school. On careful systemic review, she had no complaints, she still had a waddling gait. Her menarche occurred at age 14, menses were regular—every
twenty-eight days with a normal flow lasting three to four days. Re-examination showed a rather short stature with pronounced facial deformity and definite evidences of progressive disease in that the bony prominence of the lateral margin of the right orbit was more pronounced and there were palpable bony enlargements of the occiput with prominent frontal bosses. There were palpable bony, smooth, nodular enlargements in both humeri and over several ribs and in both tibial regions. The coxa vara was evident as before with a genu valgum more marked on the right.

Comparison x-ray films of the skull and pelvis showed evidence of progression of the disease with increased hyperostosis and cystic degeneration in the skull and pelvis.

**DISCUSSION***

Both hyperostotic and hypopostotic bone disease with normal bone structure intervening serve to distinguish this syndrome from hyperparathyroidism with bone disease. In hyperparathyroidism, hyperostotic bone lesions are most unusual and the bone disease, when present, is generalized involving a total skeletal demineralization rather than spotty disease with uninvolved normal bone structure present.

The cutaneous pigmentation in this case and segmental distribution of bone lesions involved in other cases suggest neurologic or embryologic relationships in polyostotic fibrous dysplasia. There is no such segmental pattern nor cutaneous pigmentation in hyperparathyroidism but this and precocious puberty in females are not sufficiently constant to constitute important differentials in diagnosis. The final main point in differential is the blood chemistry. The serum calcium and phosphorous levels are normal in this disease as compared to a high serum calcium and low serum phosphorous level in hyperparathyroidism. Both diseases may have a high serum alkaline phosphatase as this case shows. It will also be noted that this patient has no systemic symptoms such as those due to hypercalcemia in hyperparathyroidism, namely, muscular weakness, hypotonia, anorexia, nausea and vomiting and constipation, nor did she have evidences of hypercalciuria, hyperphosphaturia or of renal disease, which is present in over one-half of the cases of hyperparathyroidism.

Neurofibromatosis is distinguished from this disease by the fact that usually there are only a few scattered bony cystic lesions in neurofibromatosis. The cutaneous pigmentation has smooth margins like the "Coast of California" rather than the jagged margins in this case, where it resembles the "Coast of Maine" as described by Albright. The presence of multiple cutaneous fibromatous nodules and a family history of the same disorder in neurofibromatosis is in contradistinction to Albright's Syndrome. Occasionally, one may have difficulty with mild cases in distinguishing lipoid granulomatoses, solitary bone cysts, multiple myeloma, metastatic malignancy, etc., from this disease but knowledge of this syndrome and careful diagnostic study will usually differentiate these diseases.

The only known treatment is orthopedic therapy for the skeletal deformities and fractures.

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*The discussion of differential diagnosis given here is mainly taken from the book (reference four) by Albright and Reifenstein with permission of the authors.
Diagram showing localization of the bone disease in this case.

Skull x-ray showing hyperostotic bone lesions involving right orbital region with obliteration of the frontal and right maxillary sinuses.

Pelvis x-ray shows cystic changes in ilia and femora and hypostotic bone lesions. These are early changes of the "shepherds crook" deformity of femora. Normal bone structure in pubis and ischium on the left.

BIBLIOGRAPHY


