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A fragile adolescent

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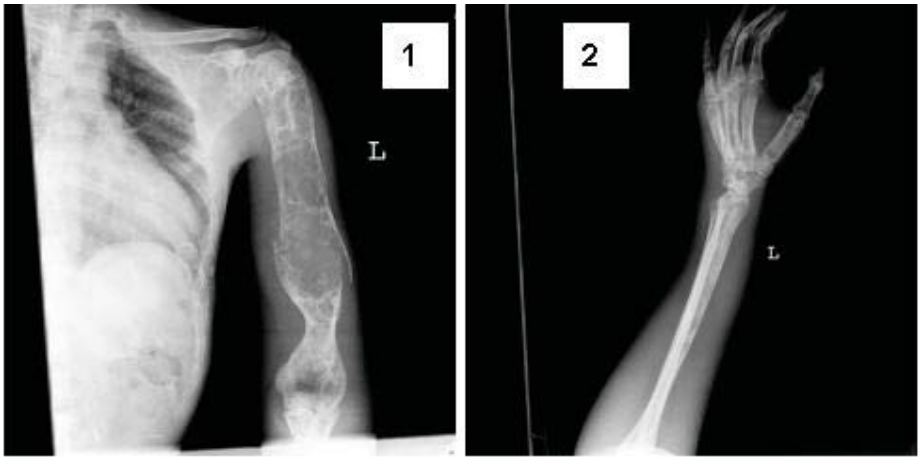
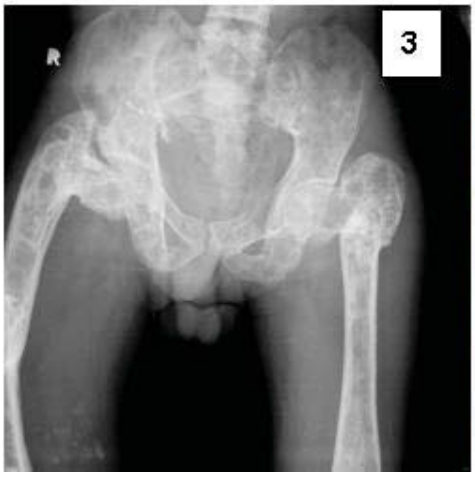
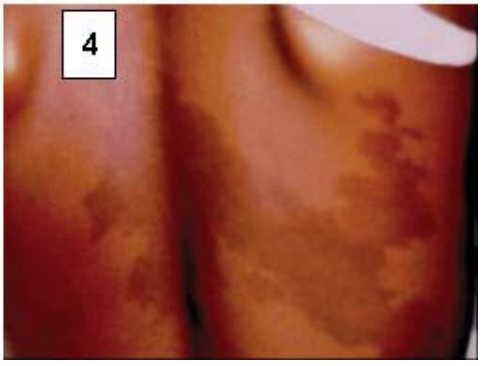
A 17-year-old boy presented with history of fracture of the left humerus sustained on minimal trauma (Figure 1). There was previous history of multiple episodes of pathological fractures with deformities since childhood. Radiological survey of the patient revealed multiple expansile lesions in various long bones (Figures 2 & 3). Physical examination revealed a dark brown hyperpigmented lesion over the upper back with irregular borders (Figure 4).







Question—*What is the diagnosis?*

Answer

The radiographs show multiple lucent lesions in the metaphysis of the long bones, with endosteal scalloping and bone expansion. This bone lesion is characteristic of fibrous dysplasia. Fibrous dysplasia is a skeletal developmental anomaly of the bone-forming mesenchyme that manifests as a defect in osteoblastic differentiation and maturation.[1]

The skin lesions in Figure 4 are called café au lait spots.[2] The presence of polyostotic (multiple sites) fibrous dysplasia and café au lait skin pigmentation confirms the diagnosis of McCune-Albright syndrome. In its classic form the triad includes autonomous endocrine hyperfunction in addition to the above two lesions. The presence of any two features among the three warrants a diagnosis of McCune-Albright syndrome (MAS).

The most common form of autonomous endocrine hyperfunction in this syndrome is precocious puberty, but affected individuals also may have hyperthyroidism, hypercortisolism, pituitary gigantism, or acromegaly.[3]

McCune-Albright syndrome is the result of a somatic mutation in the gene coding for the alpha subunit of the stimulatory G protein (Gsa).[4]

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