Tophaceous gout in thalassemia intermedia: a rare association

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A 32-year-old male presented with progressive swelling of fingers of both hands and joint pains. On physical examination, the patient had severe pallor, frontal bossing, zygomatic prominence, malocclusion of teeth and tophaceous deposits at knuckles and pinna (Fig. 1a). There was mild hepatomegaly and massive splenomegaly. On investigation, his hemoglobin was 6.1 g/dl (normal levels 13–17 g/dl), and he had microcytic normochromic anemia. He also had elevated uric acid levels of 14 mg/dl (normal levels 3.5–7 mg/dl) suggesting a hypermetabolic state. Chest and skull radiograph showed extramedullary hematopoiesis; skull radiograph revealed sun-ray appearance. Plain radiograph of the hands showed erosion of few phalanges, soft tissue swelling and 'punched-out' lytic lesions with sclerotic margins and overhanging bony edges (Martel’s sign) [1] (Fig. 1b). Joint fluid aspiration revealed needle-shaped birefringent monosodium urate crystals. A genotypic diagnosis of double heterozygous

Figure 1: (a) Tophaceous deposits at pinna and (b) Martel’s sign.
thalassemia with hemoglobin E trait and phenotypic diagnosis of thalassemia intermedia with secondary chronic tophaceous gout was made. Crystal proven gout in a patient with thalassemia intermedia is a rare but important association.

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