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IMAGES IN PEDIATRIC ENDOCRINOLOGY

Short Stature and Metaphyeal Dysplasia Due to Cartilage-Hair Hypoplasia

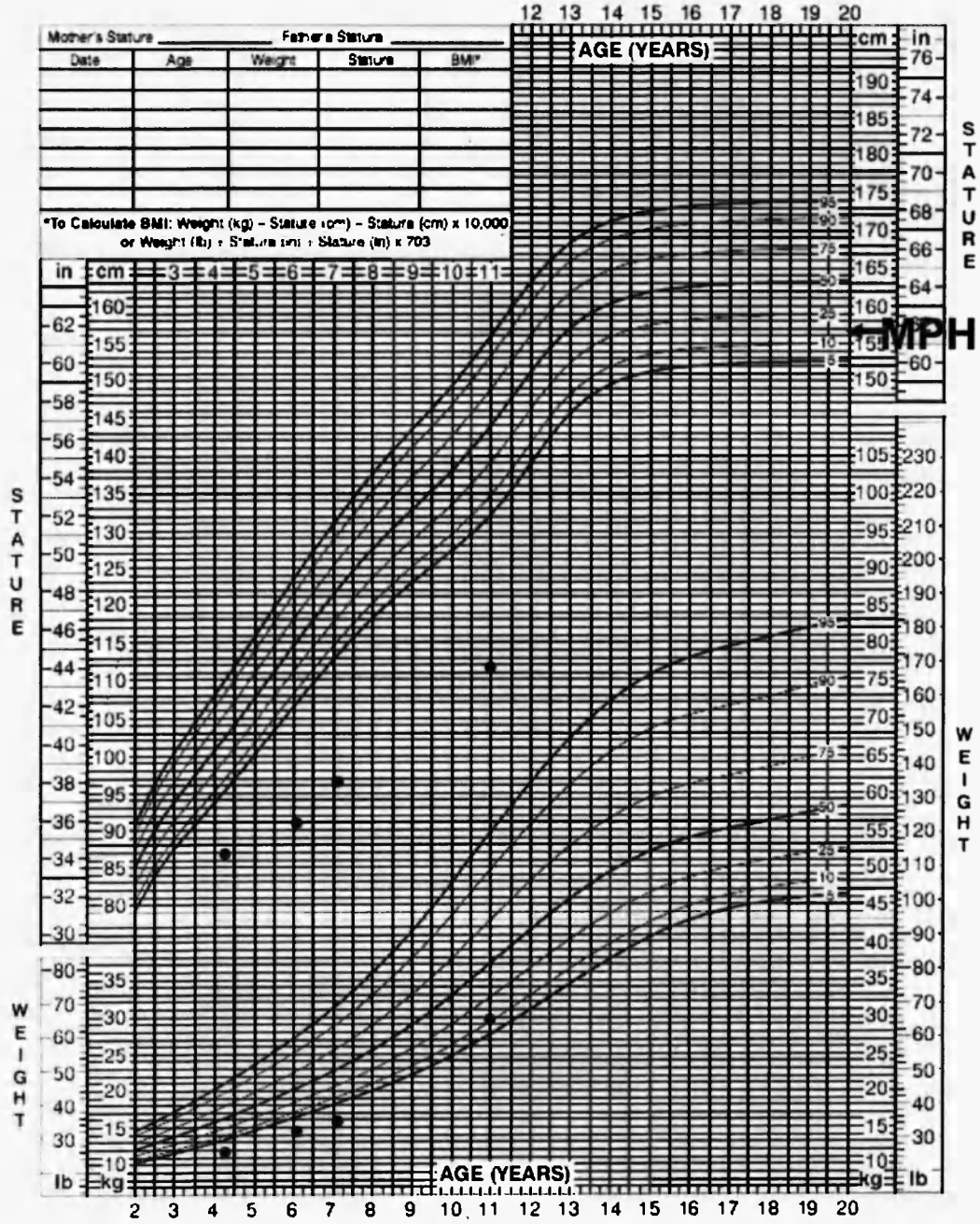
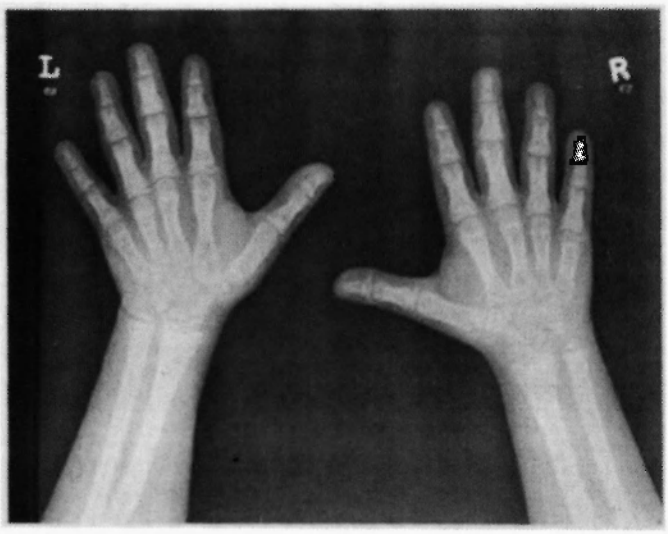
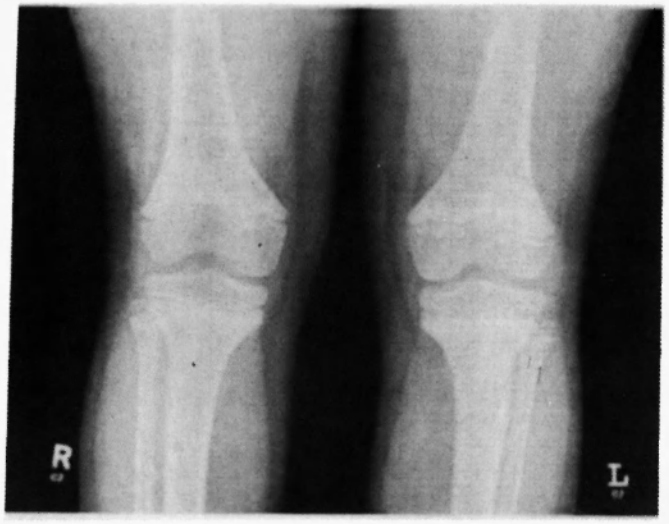


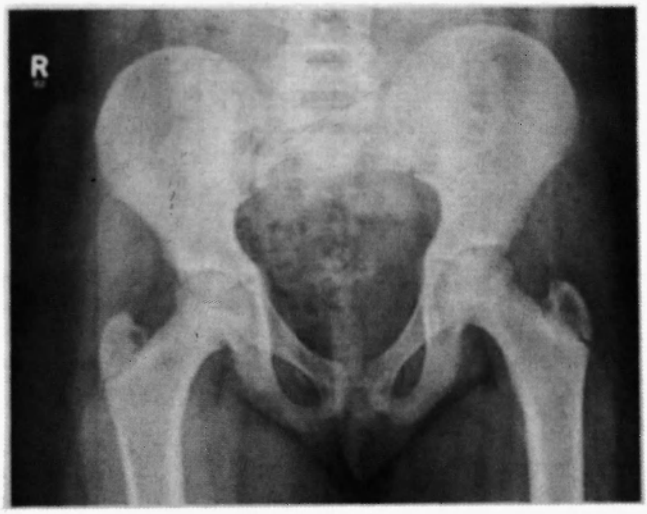
Fig. 1: Height and weight-for-age demonstrating severe and progressive growth failure. MPH = mid-parental height.



(a)



(b)



(c)

Fig. 2: Radiographs of (a) hands and distal forearms showing metaphyseal irregularities and sclerosis of the distal radii and ulnae and mildly cone-shaped epiphyses in the phalanges. Radiographs of (b) knees and (c) hips showing metaphyseal irregularities and sclerosis. The milder metaphyseal changes in the proximal femora compared to those observed in the knees are characteristic for cartilage-hair hypoplasia.

An 11 year-old presented with severe short stature and ligamentous laxity. Her height was 114.7 cm (height SDS -4.17) with mid-parental height between the 10^{th} and 25^{th} percentiles. An X-ray of the wrist performed for bone age assessment showed metaphyseal irregularities and mildly cone-shaped epiphyses. A subsequent skeletal survey revealed generalized metaphyseal dysplasia, with changes most prominent in the metaphyses of the knee. The radiographic findings were suggestive of cartilage-hair hypoplasia (CHH) and mutation analysis of the *RMRP* gene was initiated. Two pathogenic *RMRP* mutations (70A→G and 193G→A) were identified confirming the diagnosis of CHH. The patient did not have other clinical features that may be observed in this syndrome, such as fine/sparse hair, deficient cellular immunity, deficient erythrocyte production or intestinal manifestations such as Hirschsprung's disease. However, the classical, full-blown clinical picture is present in only a fraction of individuals and short stature with metaphyseal dysplasia is the single most consistent feature of CHH. A predisposition to malignancies has been reported. The *RMRP* gene on chromosome 9p21-p13 encodes for the RNA component of the ribonuclease mitochondrial RNA processing complex involved in multiple cellular processes.

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