History
Patient with dysmorphic features.

Diagnosis
Chondrodysplasia punctata non rhizomelic type

Discussion
There are three types of chondrodysplasia punctata; rhizomelic type, conradi hunerman and brachytelephalangic type. They have in common stippled epiphyses (punctate calcifications in epiphyses).

Findings in the rhizomelic type include coronal vertebral body clefting and anteriorly rounded vertebral bodies, stippling of extremity epiphyses and symmetric shortening of the humeri and femurs with less shortening of other long bones. Life expectancy is reduced.

In the conradi type; diffuse spine stippling and mildly symmetric or asymmetric extremity shortening with diffuse stippling of epiphyses. Stippling resolves during infancy. There is mild mid face hypoplasia and a high arched palate. Life expectancy is normal.

Brachytelephalangic type is far less common and patients will have severe midface hypoplasia and short hands and feet, with deformed hypoplastic proximal phalanx in the second digit in the hand and first metatarsal in the foot.

Findings

Reference
Caffeys; pp. 2637-2638.
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