Wolf-Hirschhorn syndrome: diagnosis using hand radiograph performed for bone age

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A boy aged 4 years 10 months with growth and weight retardation was admitted to our hospital. He presented with generalized hypotonia, seizures, impaired gait, peculiar facies (frontal bossing, epicanthus, irregularly shaped and low-set ears), hypospadias with undescended testes and skeletal anomalies (bilateral clubfoot, fourth-toe clinodactyly).

A bone age radiograph of the hand (Fig. 1) showed severely delayed skeletal maturation with only two carpal centres visible; basal pseudoepiphyses of the 2nd, 4th and 5th metacarpals; distal pseudoepiphysis of the 1st metacarpal; and a peculiar tongue-joint-like malformation of the distal aspects of the proximal and middle phalanges (Fig. 1).

Whilst delayed bone age can be considered a typical radiological finding for 4p- deletion or Wolf-Hirschhorn syndrome (WHS) [1], features such as tongue-joint malformations and multiple basal pseudoepiphyses are not as well known [2].

Further radiological signs of WHS include skull malformation, skeletal anomalies such as fused vertebrae, underdevelopment of the cervical spine, small pelvis with underdeveloped or absent pubic bones, “bottle opener” shaped clavicles and anomalous sternal ossification [1, 2].

Fig. 1 Delayed bone age; basal pseudoepiphyses of the 2nd, 4th and 5th metacarpals; distal pseudoepiphysis of the 1st metacarpal bone; and tongue-joint like malformation of the distal ends of the proximal and middle phalanges

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References

1. Battaglia A, Carey JC, Cederholm P et al (1999) Natural history of Wolf-Hirschhorn syndrome: experience with 15 cases. *Pediatrics* 103:830–836
2. Franceschini P, Grassi E, Marchese GS (1971) Main radiological signs of the 4p- syndrome. *Ann Radiol (Paris)* 14:335–340