Deformities of the elbow in achondroplasia

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Lack of full extension of the elbow is a common abnormality in patients with achondroplasia. We studied 23 patients (41 elbows) clinically and radiologically. Extension of the elbow was assessed clinically and the angle of posterior bowing of the distal humerus was measured from lateral radiographs.

There was limited extension of the elbow in 28 (68.3%) and the mean loss of extension was 13.1°. Posterior bowing of the humerus was seen in all elbows with a mean angle of 17.0°. There was a positive correlation between these two measurements. Posterior bowing greater than 20° caused a loss of full elbow extension. Posterior dislocation of the radial head was seen in nine elbows (22.0%). The mean loss of extension of the elbows was 28.7° which was significantly greater than that of these elbows in which the head was not dislocated (8.7°), although posterior bowing was not significantly different between these two groups (19.3° and 16.3°).

Posterior bowing of the distal humerus is a principal cause of loss of extension of the elbow. Posterior dislocation of the radial head causes further limitation of movement in the more severely affected joints.

Achondroplasia is one of the most common forms of osteochondrodysplasia with an incidence of 1 in 26 000 live births.1 It is characterised by rhizomelic, short-limbed dwarfism, relative macrocephaly, frontal bossing, a depressed nasal bridge, bowing of the lower limbs, and an increased lumbar lordosis. It is caused by mutations in the gene which encodes the fibroblast growth factor receptor 3 (FGFR3)2-4 and is transmitted as a fully penetrant autosomal dominant trait. Most cases are sporadic. The clinical features and surgical implications of deformities of the spine and lower limbs have been well described.5 By contrast, deformities of the upper limb have not been fully investigated. These include characteristic shortness of the humerus, trident hands, and variation in the length of the fingers. Lack of full extension of the elbow seems to be the earliest clinical manifestation and one of the most common abnormalities of the upper limb.

A flexion deformity of the distal humerus is commonly seen on radiographs of patients with achondroplasia and other short-limbed skeletal dysplasias which are associated with loss of extension of the elbow. Subluxation or dislocation of the abnormally-shaped head of the radius is also seen in some patients (Fig. 1). These bony abnormalities may be related to loss of extension, but to what extent has not been investigated. We have attempted to evaluate the prevalence of limited elbow extension and associated radiological abnormalities in patients with achondroplasia.

Patients and Methods

We studied 23 patients, 11 males and 12 females with a mean age of 11 years 8 months (1 year 7 months to 39 years 11 months). We excluded patients with other short-limbed skeletal dysplasias associated with extension deformity such as pseudoachondroplasia or hypochondroplasia. Both elbows were assessed clinically and radiologically. Five were excluded because adequate radiographs could not be obtained, leaving 41 elbows in the study. Extension was estimated clinically with the elbow flexed to about 90°. The flexion deformity of the distal humerus was estimated by measuring the posterior bowing angle. This was defined as the angle formed by lines drawn parallel to the mid-diaphyseal and distal diaphyseal axes (Fig. 2). Dislocation of the head of the radius was also assessed on the lateral radiographs. All measurements were made by the same investigator (HK).

Statistical analysis. Spearman’s rank correlation test was
Results

Loss of extension was present in 28 elbows (68.3%) and had a mean value of 13.1° (0 to 34). The mean age of these patients was 12.7 years which was higher than that of those without a deficit of extension (10.8 years). This was not statistically significant. Posterior bowing of the humerus with a mean value of 17.0° (10 to 28) was seen in all elbows. There was a positive correlation between loss of extension and posterior bowing ($r = 0.639$; $p < 0.0001$) in elbows without dislocation of the head of the radius. All elbows with bowing of more than 20° failed to extend fully.

Posterior dislocation of the head of the radius was seen in nine elbows (22.0%). All had loss of full extension with a mean deficit of 28.7° (15 to 34) which was greater than the mean angle of posterior bowing of 19.3° (14 to 28). In those without dislocation, the mean loss of extension was 8.7° (0 to 29) which was significantly smaller than the mean posterior bowing of 16.3° (10 to 26), but where dislocation was present loss of extension was significantly greater ($p < 0.01$), although posterior bowing was not nota-
bly different. Unilateral dislocation of the head of the radius was seen in two patients and extension of the elbow in both was more limited on the affected side although posterior bowing was not markedly different. There was a different correlation between loss of extension of the elbow and the angle of posterior bowing in patients in whom the head of the radius was not dislocated (\( r = 0.691 \)) (Fig. 3b).

Elbows in which the head of the radius was dislocated were also examined using fluoroscopy in both the flexed and extended positions in order to establish how the range of elbow movement was influenced. The head showed a prominent posterior dislocation in the flexed position (Fig. 4a) and was transferred anteriorly as the elbow was extended until it appeared to impinge and block extension (Fig. 4b).

**Discussion**

Deformities of the elbow are particularly significant diagnostically in young children with achondroplasia. Bailey reviewed the clinical and radiological findings in 41 patients and described various deformities of the upper limb including loss of extension, limited supination or pronation, a prominent radial head, a short ulna, and cubitus varus. A deficit in extension was the most common, being present in 38 of the 41 patients (92.7%). Only three patients had full extension. Bailey also observed...
that loss of extension increased with age. The number of elbows with this deformity in our series was lower (68.3%) and is probably a reflection of the relative youth of our patients. Even the 13 elbows without an extension deficit may represent a mild deformity since most normal adolescents and infants have some hyperextension of the joint.

Posterior bowing of the distal humerus was a consistent radiological abnormality and when greater than 20° resulted in loss of extension. Restriction of elbow extension, even when shoulder function is normal, may cause functional impairment of a shortened arm. Recently, surgical lengthening of the humerus has been performed to improve the function of the arm and the activities of daily living in patients with bilateral short arms. When lengthening the humerus of a patient with achondroplasia the flexion deformity should be taken into account and should be corrected simultaneously.

Radiological abnormalities of the head of the radius were difficult to evaluate in many cases. Bailey showed that the frequency of subluxation or dislocation of the head was 11.0% in patients with achondroplasia. He stated that deformities of the head, with or without dislocation, were normally the cause of loss of extension. Our study provides further evidence to link dislocation with more severe loss of full extension. The hypoplastic capitellum seen in achondroplasia suggests that the dislocation is congenital (Fig. 5). A short ulna in the forearms and a longer fibula in the lower limbs are common radiological findings probably as a result of unequal growth rates. Overgrowth of the radius, when associated with a reduced length of the ulna, may result in congenital dislocation of the head.

Developmental delay and generalised joint laxity associated with muscle weakness are common clinical features in infants with achondroplasia. The significantly smaller defect in extension in the presence of posterior bowing in those patients without dislocation suggests that the soft-tissue structures around the elbow may be lax, as is seen in other joints in achondroplasia. In younger patients these lax soft tissues are likely to compensate for the curved humerus in elbow extension. Conversely, the higher rate of elbows with limited extension in older patients may be due to relatively tight soft tissues.

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References