The 'chef's hat' appearance of the femoral head in cleidocranial dysplasia

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Cleidocranial dysplasia (CCD) is inherited as an autosomal dominant disorder characterised by failure of membranous ossification. The condition is due to a mutation of the *cbfa1* gene on chromosome 6 which has a role in the development of osteoblasts from the mesenchymal cells. In their growing years, these patients have an unusual shape of the femoral head reminiscent of a 'chef's hat'. In order to confirm the consistency of this sign, we have reviewed the radiographs of 28 patients with CCD. All except three had this appearance. The sign was also seen in patients with coxa vara associated with a variety of other conditions. The chef's hat sign may occur secondary to the particular mechanical environment created by coxa vara as well as abnormal cellular function in patients with CCD. Although coxa vara has some influence on the shape of the femoral head, it is not entirely responsible for its morphology since it was present in only six of the 28 patients with CCD.

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Cleidocranial dysplasia (CCD) is an autosomal dominant disorder. It shows various manifestations of failure of membranous ossification such as absence or hypoplasia of the clavicles, open fontanelles, wormian bones, defects of ossification of the pubic bones and supernumerary teeth.¹ Although many patients show coxa vara, some have a

normal neck-shaft angle and a few coxa valga.¹ A review of anteroposterior (AP) radiographs of the pelvis has shown that almost all have an unusual appearance of the proximal femur with elongation of the head, giving the appearance of a 'chef's hat', associated with shortening of the metaphyseal portion of the neck. We therefore carried out a retrospective study to assess the shape of the head of the femur in these patients.

Patients and Methods

The patients reviewed had been diagnosed as having CCD and had open proximal femoral physes. The diagnosis was based on either clinical and/or radiological evidence of absence or hypoplasia of the clavicle, supernumerary teeth, late closure of the fontanelles and delayed ossification of the pubis. A total of 28 patients was reviewed retrospectively (Table I). They had been seen at the Shriners Hospital, Portland and five other Shriners Hospitals within the USA in Philadelphia, Spokane, Springfield, St Louis, and the Twin Cities. Patients with scoliosis attending the Portland Shriners Hospital who had appropriate pelvic radiographs and were in the same age range, were selected as a control group. None of these 25 control patients had hip disease or related symptoms. Radiographs of the proximal femora of six patients with coxa vara, but without CCD, were also reviewed to assess the shape of the femoral head.

The presence or absence of a 'chef's hat' appearance of the head was assessed. The Hilgenreiner-proximal femoral physal line (HP) angle was used as a measure of coxa vara. To assess the shape of the femoral head, the radiograph of each proximal femur was traced on to a transparent graphic sheet on which the diameter of the proximal femoral physis (PD), the epiphyseal heights at the mid ($EH_{1/2}$), 1/4 medial ($EH_{1/4}$) and 1/4 lateral ($EML_{1/4}$) points of the physal line, and the epiphyseal diameters at the 1/3 ($ED_{1/3}$), 1/2 ($ED_{1/2}$) and 2/3 ($ED_{2/3}$) of $EH_{1/2}$ were measured (Fig. 1). The modified epiphyseal index (mEI) was calculated as the ratio of epiphyseal height ($EH_{1/2}$) relative to the epiphyseal diameter ($ED_{1/2}$). All tracings and measurements were done by one reviewer (SA). The measurements of ten randomly selected hips were repeated three times on separate days to assess reliability.

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Table I. The clinical and radiological findings for the 28 patients diagnosed with CCD

Case	Chef's hat	Pubic os def	Clavicle involvement	Supernumerary teeth	Wormian bones	Open fontanelles	Other*
1	+	+	+	+	+	+	Increase in height of proximal humeral epiphysis, horizontal acetabulum
2	+	+	+	+	+	-	Horizontal acetabulum
3	+	+	+	+	+	+	Horizontal acetabulum, elongated proximal humeral epiphysis, bilateral genu valgum
4	+	+	+	NA	+	-	Increase in height of proximal humeral epiphysis
5	+	+	+	+	NA	NA	Horizontal acetabulum, bilateral coxa vara→ ITOs, increase in height of proximal humeral epiphysis
6	+	+	+	+	NA	NA	Bilateral acetabular convexity
7	+	+	NA	NA	NA	NA	Bilateral coxa vara
8	+	+	NA	NA	NA	NA	-
9	+	+	+	+	NA	NA	Increase in height of proximal humeral epiphysis, pseudoepiphysis in the first distal and proximal second, third, fourth and fifth proximal metacarpals
10	-	+	+	NA	NA	NA	Bilateral coxa valga
11	+	+	+	+	NA	NA	Increase in height of proximal humeral epiphysis
12	+	+	+	NA	NA	NA	-
13	+	+	NA	NA	NA	NA	Bilateral coxa vara→ ITOs, bilateral ankle valgus→ medial physal stapling
14	+	+	+	NA	+	+	Increase in height of proximal humeral epiphysis
15	+	+	NA	NA	NA	NA	-
16	-	+	NA	NA	NA	NA	Bilateral coxa valga, acetabular dysplasia, subluxation
17	+	+	NA	NA	NA	NA	Bilateral convex acetabulum
18	+	+	+	+	+	NA	Pseudoepiphysis in the proximal second, third, fourth and fifth proximal metacarpals
19	+	+	+	NA	NA	NA	Increase in height of proximal humeral epiphysis, absence of pseudoepiphysis in the proximal second, third, fourth and fifth proximal metacarpals
20	+	+	+	+	NA	NA	Increase in height of proximal humeral epiphysis
21	+	+	+	NA	NA	+	Increase in height of proximal humeral epiphysis, pseudoepiphysis in the proximal second, third, fourth and fifth proximal metacarpals, bilateral coxa vara→ R ITO
22	+	+	+	+	NA	NA	Pseudoepiphysis in the first distal and second proximal metatarsal, bilateral convex acetabulum, scoliosis
23	+	+	+	NA	NA	NA	-
24	+	+	+	+	+	NA	-
25	+	+	NA	+	+	+	Horizontal acetabulum, scoliosis
26	+	+	+	+	+	-	Horizontal convex acetabulum, bilateral coxa vara
27	+	+	+	+	+	+	Increase in height of proximal humeral epiphysis
28	-	+	+	+	+	-	Bilateral coxa valga

* ITO, intertrochanteric osteotomy

Fig. 1

Diagram of PD, ED_{1/3}, ED_{1/2}, ED_{2/3}, EHM_{1/4}, EHL_{1/4}, EH_{1/2} on the proximal femoral epiphysis.

Differences between the eight radiological measurements and the mEI in the control, CCD and coxa vara groups were determined using a one-way analysis of variance (ANOVA) with the age of the patient used as a covariant. If global statistical differences were identified, Duncan's multiple comparison procedure was used to determine differences between the groups.

The reliability of the radiological measurements was evaluated using the intraclass correlation coefficient.^{2,3} This index gave both the degree of correspondence and agreement between the measurements made by the same physician on three separate days.

All statistical analyses were completed using SAS statistical software (SAS Inc, Cary, North Carolina) at a level of significance of $p = 0.05$. Based on the sample size and variability in measurements, the statistical power was greater than 70%.

Fig. 2

AP pelvic radiograph of a patient with CCD showing the 'chef's hat sign' in both proximal femora.

Results

Of the 28 patients with CCD 16 were girls and 12 were boys, with a mean age of 6.59 ± 2.75 years (2.17 to 13.17). The control group consisted of 25 patients, 17 girls and eight boys. Four had congenital scoliosis and 21 juvenile or adolescent scoliosis. The mean age of the patients in the control group was 6.50 ± 2.66 years (2 to 13). There were six patients in the coxa vara group (11 hips). Congenital coxa vara was present in two, congenital short femur in

one, proximal focal femoral deficiency in one, Schmid-type metaphyseal dysplasia in one and multiple epiphyseal dysplasia in one. All were boys with a mean age of 5.36 ± 2.08 years. Defects of ossification of the pubic bone were seen in all patients with CCD. The head of the femur resembles a 'chef's hat' when the height of the epiphysis is greater than its diameter. While the epiphysis maintains its dome shape in its upper portion it appears compressed circumferentially and elongated at its base (Fig. 2). All the patients except three with coxa vara, showed a chef's hat sign.

A summary of the radiological measurements is given in Table II. The patients with a higher HP angle (i.e., coxa vara) had a taller epiphyseal height and a lower mEI value. The only patients who showed a shorter height of the epiphysis relative to the diameter were three with coxa valga.

The intraobserver correlation coefficients for the radiological measurements of hip geometry are shown in Table III. The least reliable measurement was the medial epiphyseal height where definition of the edge of the femoral head is often obscured by radiological shadows from the pelvis and overlying soft tissues. Figure 3 depicts the shape of the femoral head for the CCD patients and control group with 95% confidence intervals.

The mean mEI in the group with coxa vara associated with diverse aetiologies was 1.22 ± 0.22 . When this was compared with the CCD and control groups, there was a significant difference between those with coxa vara and the controls ($p = 0.0001$). The mEI values for the coxa vara and CCD groups were not statistically different ($p = 0.33$). The shape of the femoral head in the group of patients with coxa vara of diverse origin resembled that in CCD (Fig. 4), although only 12 of 56 femora in the group with CCD had coxa vara (HP angle $>45^\circ$).

Table II. Summary of the radiological measurements ($\pm$ sd) and the age of the patients according to the various diagnostic groups

Parameter	CCD (n = 56 hips)	Coxa vara (n = 11 hips)	Control group (n = 50 hips)	Statistics (p value)
Age (years)	6.59 ± 2.75	5.36 ± 2.08	6.50 ± 2.66	CCD = coxa vara = control (0.8450)
HP angle (degrees)	23.64 ± 29.16	60.82 ± 19.41	17.78 ± 9.96	Coxa vara > CCD, control (0.0001)
PD (mm)	24.39 ± 7.77	24.09 ± 7.99	26.48 ± 7.24	Control > CCD, coxa vara (0.0315)
EH _{1/2} (mm)	19.64 ± 4.27	20.18 ± 4.56	14.12 ± 3.01	CCD, coxa vara > control (0.0001)
EHM _{1/4} (mm)	17.34 ± 4.65	18.64 ± 3.50	11.78 ± 2.81	CCD, coxa vara > control (0.0001)
EHL _{1/4} (mm)	18.77 ± 4.16	19.18 ± 4.33	13.74 ± 3.33	CCD, coxa vara > control (0.0001)
ED _{1/3} (mm)	26.32 ± 10.28	24.27 ± 7.64	26.16 ± 17.00	CCD = coxa vara = control (0.7365)
ED _{1/2} (mm)	26.59 ± 8.45	24.73 ± 7.46	24.48 ± 5.54	CCD > coxa vara, control (0.0420)
ED _{2/3} (mm)	23.37 ± 6.19	21.91 ± 6.31	19.78 ± 4.91	CCD, coxa vara > control (0.0001)
mEI	1.35 ± 0.315	1.22 ± 0.216	1.74 ± 0.244	Control > CCD, coxa vara (0.0001)

Table III. Intraobserver correlation coefficients for the radiological measurements

	PD	EH _{1/2}	EHM _{1/4}	EH _{1/4}	ED _{1/3}	ED _{1/2}	ED _{2/3}	mEI
Intraobserver correlation coefficients	0.9850	0.9691	0.4139	0.8643	0.9045	0.8105	0.9274	0.8924

Fig. 3

Profiles of the femoral head of patients with CCD and the control group.

Discussion

In 1898, Marie and Sainton⁴ reported four patients with cleidocranial dysostosis. They described the classical findings of delay of ossification and closure of the fontanelles, disturbance of dentition, hypoplasia or aplasia of the clavicles, enlargement of the cranial vault and hereditary transmission. In 1973, Fauré and Maroteaux⁵ noted also that the femoral necks were usually deformed. Unilateral or bilateral coxa vara was present in approximately half their cases. They described the capital femoral ossification centre as being deformed, with a rounded appearance and tapering at its edge directed towards the neck. Jarvis and Keats⁶ reviewed 40 patients with CCD. They observed lateral notching of the proximal femoral ossification centre in three children, and noted that both the femoral neck and head showed widening. Wynne-Davies, Hall and Apley⁷ and Spranger, Langer and Widemann⁸ mentioned that the femoral head in CCD patients was abnormal in shape.

In our study, all CCD patients, except three with coxa valga, demonstrated the unusual shape of the femoral head which we have called the 'chef's hat' sign. This was confirmed by objective measurements which showed an increase in the height of the epiphysis relative to the diameter compared with normal. In six patients with coxa vara associated with a diverse group of skeletal problems, the chef's hat appearance of the femoral head was also present. As a result we suggest that the particular mechanical environment of the proximal femur induces it to develop this unusual shape. There must, however, be other

Fig. 4

AP radiograph of the pelvis of a patient with Schmid-type metaphyseal dysplasia.

factors present in CCD since the chef's hat shape also occurs in CCD patients without coxa vara. Komori et al⁹ and Otto et al¹⁰ deleted the *cbfA1* gene in mice by homologous recombination. The most pronounced effect was an absence of membranous and endochondral bone in the fetal

homozygous *cbfA1* $-/-$ mice, a condition which was always lethal. The *cbfA1* $+/-$ heterozygous mice showed hypoplasia of the clavicles, delayed development of membranous bones, and delayed ossification of cranial bones with open fontanelles and multiple wormian bones, similar to the features of CCD.¹⁰ Mundlos et al¹¹ examined the *cbfA1* gene in man. Their study showed a lack of expression of one allele of the *cbfA1* gene as the basis of CCD in man which was fully consistent with the phenotype of the *cbfA1* $+/-$ heterozygous mice except for the supernumerary teeth in human CCD. The groove of Ranvier in the periphery of the epiphyseal cartilage has a role in the appositional growth and mechanical stability of the growth plate.¹² Densely packed mesenchymal cells which are the progenitors of osteoblasts are found deep in the groove.¹³ Since the *cbfA1* gene defect may affect the ability of these mesenchymal cells to differentiate, this may be a factor in the unusual development of the proximal femur.

Although rare, the chef's hat appearance can be seen in patients with coxa vara of diverse aetiologies. When AP radiographs of this pelvis show this distinctive feature and absent ossification of the pubic symphysis, the diagnosis of CCD should be considered.

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