

## Acromesomelic Dwarfism: Report of a Family with Two Affected Siblings

S. Danda  
Shubha R. Phadke  
S.S. Agarwal

Acromesomelic dwarfism is a skeletal dysplasia characterized by disproportionate short stature with predominant involvement of the forearms and hands. Maroteaux *et al.*(1) first described this form of dwarfism. Over 36 cases have been described till date(2). Of them 3 families consisting of two siblings with definite diagnosis of acromesomelic dwarfism are described(3). Here, we report an Indian family with two affected siblings.

### Case Reports

A 12 year old boy (*Case I*) and his 3 year old sister (*Case II*) were brought to Genetics Clinic for the problem of short stature. The parents were non-consanguineous, Hindu couple from Uttar Pradesh, India. Besides these two children, the couple had two full term still births and a boy who died of dehydration in the eighth month of life. No medical records or photographs of these children were available. The parents were of normal height and proportion.

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*From the Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences Rae-Bareilly Road, Lucknow 226 014.*

*Reprint requests: Dr. Shubha R. Phadke, Assistant Professor, Department of Medical Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Rae-Bareilly Road, Lucknow 226 014.*

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*Case I:* He was the first child of the couple. Pregnancy and perinatal period was normal. Short stature was noticed at 3 years of age. At the same time deformities of elbows and forearms were noticed (*Fig. 1*). The deformities have been increasing since then. Developmental milestones were normal. There was no difficulty in vision or hearing.

At 12 years of age, height and weight were 96 cm and 20 kg, respectively, both being less than third percentile. Head circumference was 53.5 cm which was normal for the age. Ratio of upper segment to lower segment was normal (1.1). Arm span (72.5 cm) was significantly less than the height (96 cm). This shows that involvement of upper limbs was more severe than that of lower limbs.

On examination, the skull was dolicocephalic with frontal bossing and occipital prominence. He had broad, depressed nasal bridge, high arched palate and carious teeth. There was extreme shortening of upper limbs more marked in forearms and hands. There was bowing of forearm bones leading to convexity of the radial border of forearm. Terminal extension at elbows was restricted. Pronation and supination were normal. Fingers were very short. The lower limbs though short were less as compared to upper limbs. The feet were short, flat and square with relatively large great toes and proximally placed second toe bilaterally. The nails were broad and short in both hands and feet.

Movements at other joints were normal. Lumbar lordosis was exaggerated. He had pectus carinatum. Serum creatinine, calcium, phosphorus and alkaline phosphatase were normal. The radiographs showed short, stubby long bones with flaring at ends. Radius and ulna were more severely involved (*Fig. 2*). There was bowing of

radius and distal end of ulna was shorter than radius creating a gap between the distal end of ulna and carpals at the wrist. The distal radial and ulnar epiphyses were unusually large and rounded, the adjacent metaphysis were flared. The metacarpals were short, broad with flared ends. The phalanges were short with shortening most marked in distal phalanges. Similar shortening was noted in metatarsals and phalanges of feet but was less marked. The first metatarsal and phalanges of great toe were broad. Lateral view of spine showed platyspondyly. The borders of the vertebrae were regular with decreased height in the posterior part (posterior wedging). The iliac bones were hypoplastic. The clinical and radiographic features were diagnostic of acromesomelic dwarfism.

*Case II:* The younger sister of Case I was 3 years old. Short stature was noticed since 2 years of age. Her milestones were normal. Her height (62 cm) and weight (8 kg) were less than third percentile (*Fig. 1*). Head circumference was 55.5 cm (>95th percentile). The head was large and dolicocephalic with marked frontal and occipital prominence. Face was similar to that of her brother with depressed and broad nasal bridge. The wrist and ankles were broad. Shortening and deformities of forearms were obvious but were less marked as compared to her brother. Hands were short with very short fingers. Her parents refused to get her investigated. In spite of lack of radiological proof, the similarity of her clinical features with those of her brother left no doubt about her being affected with the same condition as that of her brother.

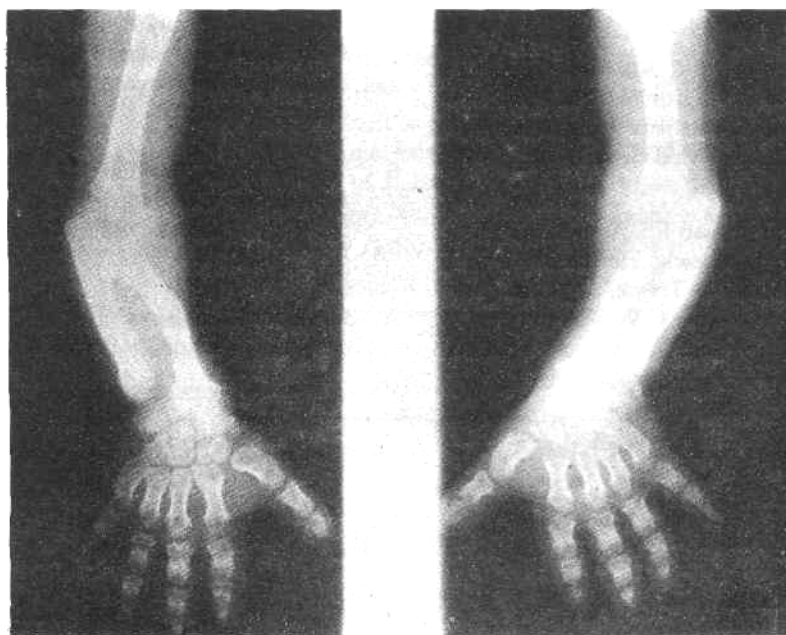
#### Discussion

The two siblings described above have characteristic clinical and radiological features of acromesomelic dwarfism. Both the sibs had marked short stature, more severely involving forearms and hands.

The various skeletal dysplasias associated with acral or acromesomelic shortening are acromicric dysplasia, Geleophysic dysplasia, Trichorhinophalangeal syndromes (Type I and II), acrodysostosis, Saldino Mainzes syndrome, Albright hereditary osteodystrophy and Acromesomelic dysplasia(4). Out of these, acromicric dysplasia and acrodysostosis have some similarities to the above cases. Acromicric dysplasia is characterized by markedly short hands and feet, dwarfism to variable degree and characteristic face with narrow palpebral fissures, short stubby nose and anteverted nostrils(5). They have limitation of finger flexion and a round shaped pseudoepiphysis at the distal end of first metacarpal(4). Absence of the above features and



*Fig. 1. Photograph showing both siblings with disproportionate shortening and deformities of elbows.*



*Fig. 2. Radiograph of upper limbs of Case 1.*

presence of forearm shortening and deformity in the present cases is against the diagnosis of acromicric dysplasia. Acrodyostosis is characterized by moderate short stature, mental retardation and brachycephaly(4). The present cases have severe short stature, normal intelligence, scaphocephaly, acromesomelic shortening and forearm deformities which are diagnostic of acromesomelic dysplasia(6). Lumbar lordosis, large first toe and dislocation of head of radius is also described in acromesomelic dysplasia(4). Our cases have all the above features.

The usual age at diagnosis is around 3 years. In familial cases, however the diagnosis may be suspected earlier in second affected child as was the case in the above family. Adult height usually ranges from 94 to 123 cms(7).

Corneal opacities have been previously

described in 2 cases(3). Our patients did not have corneal opacities. Three pairs of affected sibs are described previously(3). These families and an affected case with consanguineous parents(2,3) support an autosomal recessive mode of inheritance. Though, there was no consanguinity in the present family the parents belonged to the same caste of Kewats. The normal parents and 2 affected sibs in the above family support autosomal recessive pattern of inheritance. This communication appears to be the first report of acromesomelic dysplasia from India.

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